Artificial Intelligence for Genetic Cancer Risk Assessment in Gynecologic Oncology

A Review of the Current Landscape and Future Directions

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Abstract: Hereditary cancer syndromes are associated with up to 25% of ovarian and 5% of endometrial cancers, yet rates of genetic testing and counseling remain low. Artificial intelligence (AI) offers new opportunities to streamline risk assessment, enhance gene variant interpretation, and expand access to genetic counseling. This narrative review synthesizes current evidence on AI applications in gynecologic cancer genetic risk assessment, including chatbot-based risk assessment, natural language processing of electronic records, and machine-learning approaches to variant classification. We highlight key challenges, including data bias, privacy, and implementation barriers, and outline future directions for AI technologies in gynecologic cancer genetic risk assessment.

Key Words: cancer, genetics, artificial intelligence, hereditary breast and ovarian cancer, risk screening, gynecologic oncology

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INTRODUCTION TO GENETIC RISK ASSESSMENT FOR GYNECOLOGIC CANCERS

Genetic Risk in Gynecologic Cancers

Approximately 5% to 10% of all cancers are associated with hereditary cancer syndromes.[1] Identification of individuals who carry pathogenic variants, or mutations, in genes linked to hereditary cancer syndromes is critical for not only early detection and cancer prevention but also decisions around fertility and reproductive choices. In addition, knowledge of genetic status can facilitate predictive genetic testing for family members (cascade testing) and guide targeted cancer therapies such as poly (ADP-ribose) polymerase (PARP) inhibitors.[2] However, fewer than 20% of individuals who carry pathogenic variants that cause these syndromes are aware of their genetic status.[3–5]

Among gynecologic malignancies, hereditary cancer syndromes are typically associated with ovarian and endometrial cancers. An estimated 20% to 25% of ovarian cancers are attributable to hereditary cancer syndromes, largely driven by pathogenic variants in *BRCA1* and *BRCA2*.[6] A smaller proportion of ovarian cancer cases are linked to genes involved in homologous recombination repair (such as *RAD51C, RAD51D, BRIP1,* and *PALB2*) and mismatch repair genes associated with Lynch syndrome.[7] By contrast, 3% to 5% of endometrial cancers are attributable to hereditary cancer syndromes, with Lynch syndrome responsible for the vast majority of cases, involving mismatch repair (MMR[8]) genes (*MLH1, MSH2, MSH6,* and *PMS2*).[9,10]

Accordingly, multiple organizations—including the Centers for Disease Control and Prevention, the American College of Obstetricians and Gynecologists, Society for Gynecologic Oncology (SGO), and National Comprehensive Cancer Network (NCCN)—have emphasized the importance of genetic testing for those at high risk for carrying pathogenic variants.[8,11–13] In addition, the NCCN and SGO recommend that all individuals diagnosed with epithelial ovarian cancer undergo genetic counseling and testing.[7,12,13] For endometrial cancer, NCCN guidelines also recommend universal counseling and testing, while SGO recommends genetic testing for any individuals found to have microsatellite instability or MMR deficiency on immunohistochemistry testing of the tumor, which is recommended for all patients with endometrial cancer across guidelines.[8,12,14,15] Current screening practices comprise both history-based counseling and cascade testing to increase uptake of genetic testing in patients with hereditary cancer syndromes and their at-risk relatives.

In traditional counseling, providers review patients' personal and family histories to identify individuals meeting criteria for high risk of hereditary cancer syndrome. This conventional risk assessment relies on models such as Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation (BOADICEA), BRCAPRO, and Prediction Model for Mismatch Repair Gene Mutations 5 (PREMM5) to estimate mutation probability based on family history and clinical features.[16] While these tools are valuable, their performance is often limited by incomplete family histories and biases inherent in the populations from which they were derived. In cascade genetic testing, patients with cancer undergo genetic testing and, if found to have positive screening, disclose results ("cascade") to their at-risk relatives, who subsequently pursue their own genetic testing. However, adherence to guidelines for genetic testing remains low among patients diagnosed with gynecologic cancers, with referral rates to genetic services estimated at 13% to 53% and subsequent testing rates ranging from 5% to 92%.[17–19] Thus, the suboptimal rate of genetic testing

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among cancer patients also limits the number of at-risk relatives who undergo screening through cascade testing.

One study by Lin et al[17] reviewed 35 studies on genetic assessment of ovarian cancer patients through 2020. They reported a pooled rate of referral to genetic counseling of 39% and pooled genetic testing rate of 30%.[17] For endometrial cancer, referral rates to genetic counseling have ranged from 13% to 100%, with subsequent testing rates of 61% to 94%.[18] Studies of cascade genetic testing suggest even lower rates of uptake for at-risk relatives, with estimated genetic testing rates of 15% to 51% for first-degree relatives of individuals diagnosed with a gynecologic cancer.[4,18] A systematic review of 87 studies evaluating hereditary cancer cascade testing through 2021 reported a pooled genetic counseling rate of 48% for relatives and cascade genetic testing rate of 41%.[20]

Current Barriers to Uptake of Genetic Cancer Risk Assessment

Several barriers to uptake of genetic cancer risk assessment have been identified, from the individual to systems level. At the individual level, barriers include incomplete family histories, perceived cost of genetic testing, and limited knowledge of the genetic screening process.[21–23] In multiple dimensions, fear can also act as a powerful barrier: internalized fear of being identified as high-risk for cancer, fear of genetic discrimination both interpersonally and related to insurance, and distrust of the medical system.[24,25]

At the provider level, barriers include issues related to health literacy and subsequent comfort level discussing genetics, time constraints, and logistics of coordinating genetic screening. Primary care providers (PCPs) are ideally positioned to perform routine genetic screening for all-comers.[26] However, this requires PCPs to prioritize conversations around genetic risk, know how to identify high-risk patients, understand how to order and interpret genetic testing, and coordinate patients with follow-up care.[27–29] Although continuing medical education can address some of these barriers, its effects are less likely to be systematic, dependent on individuals' decisions to pursue these courses. For all providers, beyond PCPs, another barrier can be familiarity with—and the availability of—genetic counselors, who can help with appropriate counseling for patients and further referrals to care and resources.[27,30] The current and growing demand for counseling by certified genetic counselors far exceeds the workforce supply, a bottleneck particularly notable for patients living in rural communities, given the unequal distribution of clinical genetic services across the United States.[31,32] However, even beyond knowledge and resource gaps, time is a key barrier for providers, who are then tasked with allocating enough time within already constrained appointments to go through screening questions and subsequent counseling regarding genetic testing.[29]

At the systems level, genetic testing processing and screening tools have not been fully integrated into electronic medical record (EMR) systems.[33] Although most EMRs will contain sections for family history, standardized tools to calculate risk for genetic cancer syndromes have not been integrated into most systems. Furthermore, even if a patient overcomes the individual- and provider-level barriers, hesitation may arise due to concerns around the cost of genetic testing services.[34,35] Both patients and providers may be unsure of whether insurance will cover the cost of testing as well as subsequent genetic counseling. Out-of-pocket costs for genetic testing can range from $100 to $2000 depending on the type of genetic testing and extent of insurance coverage. Most laboratories offer out-of-pocket options for $250 or less.[36] However, Medicare does not provide coverage for presymptomatic or prediagnosis genetic testing for hereditary cancer risk.[37]

Role of Artificial Intelligence in Genetic Risk Assessment

Artificial intelligence (AI) offers a transformative opportunity in hereditary gynecologic cancer risk assessment. AI encompasses several computational techniques that identify nonlinear relationships in data.[38] For example, deep learning uses multilayered neural networks through training on extensive data sets to identify complex patterns, such as in large-scale genomics data. Natural language processing (NLP) enables automated processing of human language, with the promise to extract genetic and family history from unstructured EMR data. Through these techniques, AI systems can integrate diverse data sources —including genomics, clinical characteristics, imaging, biomarkers, and family history—into unified predictive models that create more comprehensive and individualized assessments of genetic gynecologic cancer risk.[33] Furthermore, AI holds the potential to increase equity in hereditary cancer risk assessment not only by creating automated, streamlined workflows but also through powering chatbots that can interpret complex data and offer counseling for patients. Through supplementation of in-person genetic counseling services, AI can expand cancer screening accessibility, including for patients in low-resource or rural settings where access to specialized services is more limited.[33] Thus, through the integration of multiomic data, EMR, and population data sets, AI holds promise to improve genetic risk prediction, interpretation, and accessibility in gynecologic oncology (Fig. 1).[39,40] This review synthesizes current evidence on AI applications in genetic cancer risk assessment for gynecologic cancers, identifies challenges and limitations, and highlights future directions for research and clinical practice.

METHODS

For this narrative literature review, AI was used as a MESH term on PubMed and associated with terms including "gynecologic cancer," "gynecologic oncology", "ovarian cancer," "endometrial cancer," "uterine cancer," or "cervical cancer." Results were searched through September 30, 2025. We selected published evidence about the role of AI in genetic cancer risk assessment for gynecologic cancers and summarized the key findings. Publications focusing on the role of AI in the treatment (surgical or medical) or assessment of risk recurrence in gynecologic cancers were not included. Similarly, publications focusing on breast cancer exclusively were not included.

CURRENT APPLICATIONS OF ARTIFICIAL INTELLIGENCE IN GENETIC GYNECOLOGIC CANCER RISK ASSESSMENT

Artificial Intelligence in Gynecologic Cancer Risk Prediction and Screening

Several studies have employed AI to standardize and streamline genetic screening for gynecologic cancer risk,

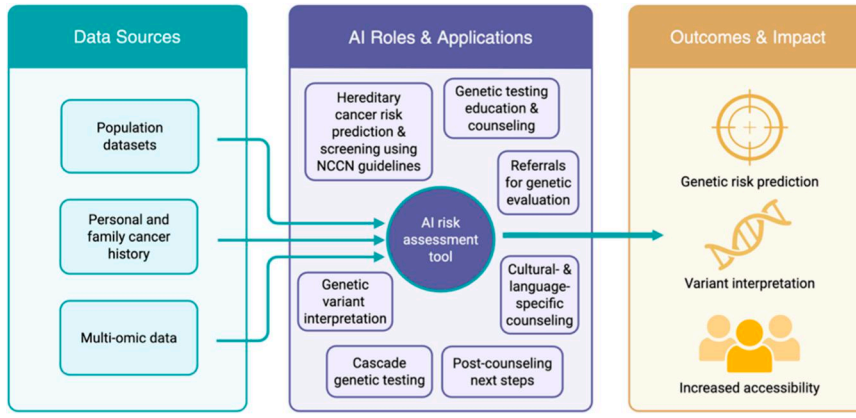

FIGURE 1. Overview of the potential applications and impacts of artificial intelligence in gynecologic cancer risk assessment. Created in BioRender. Frey, M. (2025) https://BioRender.com/0u57ea0. full color online

accounting for personal and family histories. Most of these studies evaluated the efficacy of a chatbot for risk prediction. One of the earliest studies to evaluate the efficacy of AI-based tools for hereditary cancer risk stratification was a 2016 retrospective review of 100 patients' charts from a US-based adult genetics clinic.[41] In this study, a custom AI-based tool automatically extracted clinical data from patients' EMRs (including problem lists, laboratory values, and pathology reports translated through natural language processing) to determine whether patients were deemed high-risk for hereditary cancer syndromes. Through comparison with retrospective chart review conducted by genetic counselors, the study reported 97% agreement between patients deemed high-risk by the tool and NCCN guidelines. For ovarian cancer specifically, the tool was found to have 93% agreement with a geneticist's risk assessment (n = 62/67). For uterine cancer, the agreement rate was only 50% (n = 2/4). As patients were selected from a genetics clinic, the study sample was biased towards high-risk patients, with 53% of participants having a personal history of cancer. Thus, findings from this feasibility study lacked external validity for use as a wider, population-based screening tool.

Since then, multiple studies have evaluated web-based chatbots that collect personal and family histories and automatically generate risk estimates for hereditary cancers. In a study of 15,000 patients from 28 OB/GYN clinics in the United States, 65% of invited participants completed a chatbot on personal and family cancer histories, of whom 26% met NCCN screening criteria for hereditary cancer testing.[42] Similarly, in a multicenter cohort study across 180 US clinics (not limited to OB/GYN-related care), a total of 95,166 patients were invited to use a web-based chatbot before routine appointments to assess risk for hereditary breast and ovarian cancer (HBOC), Lynch syndrome, and adenomatous polyposis syndromes.[43] Most participants were females (96.3%) and white (58.5%), with nearly half aged 40 to 60 years (45.5%). Less than 11% of users reported previously being offered genetic testing. Acceptability of the chatbot was high, with 89.4% completing the risk assessment. Among those screened, 27.2% met NCCN criteria for genetic testing—including 20.4% for HBOC, 2.4% for Lynch syndrome, and 2.7% for multiple criteria. For those patients whose health care providers could directly order genetic testing, 29% were ordered for genetic testing, among whom 5.6% were found to have pathogenic/likely pathogenic variants and 26% found to have variants of unknown significance (VUS). The chatbot did not automatically add patients' risks to their EMRs, but the tool generated composite data that could be transferred to the EMR. However, as neither of these two studies compared screening rates to routine clinical care, they were unable to assess whether the chatbot was associated with improved uptake of preventive measures.

The feasibility of automating hereditary cancer risk assessment at the population level was assessed in a study that recruited participants through online marketing campaigns across the United States.[44,45] In this study, a chatbot gathered personal and family histories, which were automatically transferred to a web-based application capable of assessing whether participants were eligible for cancer genetic consultation, in accordance with NCCN and American College of Medical Genetics and Genomics (ACMG) guidelines.[45,46] Of a total of 4915 female patients who completed the chatbot questionnaire, most were 40 to 59 years old (92.5%) and of European descent (38.6%).[44] Overall, 45.2% met either NCCN or ACMG criteria for genetic cancer consultation referral, and 39.8% met NCCN criteria for genetic testing.[45] ACMG does not provide criteria for genetic testing. Among those meeting NCCN testing criteria, 10.4% met criteria for breast, ovarian, and pancreatic cancer testing, while 28.6% fulfilled criteria for colorectal and endometrial cancer. While this study did not compare findings to screening rates per routine clinical care, only 5.2% of participants reported personal genetic testing, which may reflect the outcomes of routine history-based screening and testing per clinician judgment. Although this rate could arguably represent the comparator group, it more precisely reflects uptake of genetic testing after routine screening, and rates of genetic testing after chatbot screening were not reported in the study. In this study, the high rate of positive screening is likely due to sampling and participation bias, as recruitment methods included targeted campaigns directed towards women aged 40 to 60 with interest in cancer and directly contacting cancer support groups.

However, one study did compare uptake of cancer genetics referrals and testing between individuals identified

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through chatbot and through standard-of-care models.[47,48] This randomized controlled trial by Kaphingst et al[47] enrolled 3073 patients at 2 US health care systems, who were English or Spanish-speaking with no personal cancer history or prior cancer genetic services. Participants in the intervention group (50.6%) completed a pre-test genetics education chat, while those in the control group (49.4%) completed a pre-test appointment with a certified genetic counselor. Overall, 72.9% of participants were female, 1.3% were Spanish-speaking, and 75% were white, with a mean age of 43.8 years. The study found no significant differences in completion of pre-test cancer genetic services or in completion of cancer genetic testing between the two study arms. These equivocal findings support the use of chatbots for cancer genetics services, especially considering the scalability and cost-effectiveness of chatbots.[49] Of note, even patients in the chatbot intervention arm were contacted by a genetic counselor to confirm their genetic testing decision. Secondary analysis from this data set found that patients with greater socioeconomic vulnerability —regardless of study arm—were less likely to initiate or complete pre-test genetic services and also less likely to have genetic testing ordered.[50] Thus, use of a chatbot alone cannot mitigate socioeconomic barriers to genetic testing care, and further work is required to optimize chatbots' acceptability for more vulnerable populations.

Outside of the United States, Sato et al[51,52] have evaluated the feasibility of a chatbot integrated into the interface of an existing social media platform for the evaluation of HBOC risk in Japan. For the 11 participants in their study, the chatbot's assessment of eligibility for *BRCA1/2* testing was compared with assessment by a certified genetic counselor.[52] For 3 (27%) participants, discrepancies were noted between the histories obtained by chatbot and counselor, and families histories were incomplete for 6 (54%) participants. Identified reasons for these disparities included participant omission, incomplete questions from the chatbot, and communication errors with the chatbot. While the study demonstrated clinical feasibility for the chatbot, it also highlighted the need to further revise chatbot language to obtain more accurate histories.

Beyond chatbots, NLP models have been applied to patients' EMRs to improve the identification of patients eligible for cancer genetics services.[53] Missing data in family health history is a major barrier to accurately identifying eligible patients, with significant disparities in completeness by sex, race, and language.[54] To address this barrier, one study applied an NLP component to EMR data from patients (n = 120,007) at a tertiary academic medical center in the United States, where a rule-based algorithm was already in place to identify patients meeting NCCN criteria based on structured family history data.[55] The authors found that enhancing the structured data set with NLP, which drew from free-text comments in the EMR, increased the detection rate of eligible patients by 1.9%. Among the new patients identified through NLP enhancement, 28.3% were eligible for genetic testing through family history of ovarian cancer. However, the study population was limited to one health care system in one US state (Utah), using a specific EMR, which does not reflect national distributions of ethnicity or usability of free-text content in EMRs.

Artificial Intelligence for Interpretation of Genetic Variants

A growing field of research within AI applications in oncology is the use of AI to interpret genetic variants, particularly VUS. This field expands beyond hereditary cancer syndromes and investigates individual-level risk for cancers based on genetic profiles, leveraging genomics databases.[56] Currently, the ACMG and Association for Molecular Pathology (AMP) have created a joint standard for interpreting and classifying variants that draws from population, computational, functional (in vivo/in vitro/in silico), segregational, and inheritance pattern data.[57]

One study utilized a database of over one million variants derived from next-generation sequencing (NGS) of 10,116 patients from a cancer center in Australia to generate AI models that assist in the interpretation of genetic variants.[58] Variants were analyzed by trained genomic scientists, who identified variants appropriate for reporting. These curated and annotated variants were compiled into a database used to generate machine learning (ML) models that identified subsequent patients who presented with the same variants with their respective clinical contexts. The models performed well, with precision recall area under the curve (AUC) ranging from 0.907 to 0.995. The AI models streamlined future analyses such that only novel variants required manual curation by experts. Thus, in this application, ML models facilitated more streamlined, thorough analyses of variants within patients' clinical contexts, alongside manual expert curation. Through this more efficient processing of genomic data, the ML model potentially enables upscaling of genetics services. Of note, this study did not provide discrete data by tumor type, so the validity for gynecologic cancers specifically could not be determined. In addition, generalizability of the models was limited by the sourcing of all data from a single laboratory.

Although no identified studies focused on AI models for variant interpretation in gynecologic cancers specifically, several have evaluated *BRCA1/2* variants in the breast cancer context. The ACMG/AMP standard has been employed by the international Evidence-Based Network for the Interpretation of Germline Mutant Alleles (ENIGMA) consortium, which classifies *BRCA1/2* variants.[59] So far, the ENIGMA consortium has reviewed at least 3335 *BRCA1* variants, but there are over 31,000 *BRCA1* variants that have not yet been reviewed for classification.[59,60] One study developed a more comprehensive, AI-based prediction model to aid in interpretation of the unclassified variants and to test the model on a Qatari breast cancer patient cohort. The prediction model was trained and tested on the 3335 variants reviewed by the ENIGMA consortium, with 99.7% to 99.9% accuracy. The prediction model was then applied to the 31,058 unreviewed variants in the BRCA Exchange database. In the Qatari population, only 12 of the 36 *BRCA1* variants were already classified by ENIGMA. The *BRCA1* model was used to predict the pathogenicity of unreviewed or new variants, all of which were classified as benign. Similarly, another study used two different AI methods—fuzzy logic and neural network systems—to assign breast cancer risk scores to individuals with *BRCA1/2* VUS using patients' other clinical characteristics.[61] These AI models were trained on 268 patients from 2 Turkish cancer cohorts and tested on 12 patients, with 95.5% accuracy. As these studies drew upon data regarding breast cancer outcomes, their interpretation in the gynecologic context is limited. However, they offer a template for parallel efforts to apply AI techniques in interpreting VUS in *BRCA1/2* and other genes relevant for gynecologic cancers.

Artificial Intelligence in Genetic Counseling

Aside from screening for and estimating genetic cancer risk, chatbots have also been integrated into routine tasks in the genetic screening cascade from pre-test counseling to obtaining informed consent to delivery of negative results and cascade testing.[62–64] Integrating AI into genetic counseling has aimed to standardize genetic counseling, improve patients' understanding of their results and individual risks, and to address the gap between the growing demand for genetic counseling and the limited supply of certified counselors.[65]

In one of the earlier chatbot studies, Schmidlen et al focused on the use of a chatbot designed to gather genetic testing consent from participants in an existing research project that couples genomics data with EMR at an integrated rural health care system in the United States.[66] In this study, participants were notified of their genetic testing results by a genetic counselor and offered free genetic counseling, with subsequent 1 and 6-month follow-ups with the genetic counselor. Given the extensive direct counseling already integrated into the project, the qualitative study focused on the use of chatbots in the initial consent process. The chatbot allowed participants to tailor the amount of detail they wanted to receive on different topics (eg, goals, risks, benefits) during the consent process. Among the 62 participants, overall support for the consent chatbot was high, with participants appreciating the ease of use and ability to determine the speed and content of conversation. Further, they noted the chatbot process to be more thorough and informative than prior in-person consenting experiences. While the feedback was overall favorable, participants also emphasized the importance of continuing the in-person counseling option. For follow-up, the chatbot also offered several reminders of suggested actions following receipt of their results, including an option to schedule a counseling visit. In addition, a cascade chatbot was developed to enable participants to share their testing results with family members through chatbot link. The cascade chatbot described the proband's result, risks, and recommended management, with the option for relatives to request a genetic counseling visit. The cascade chatbot was favorably received for its ability to answer key questions for relatives, but several participants noted that the chatbot would be less appropriate for certain relatives, based on characteristics such as their age, attitudes towards medicine, or familiarity with the proband. Participants also expressed concerns regarding the security and privacy of information in the chatbot link.

In a systematic review of 7 US-based studies that evaluated chatbot use for genetic cancer risk assessment through 2022, 4 studies described chatbot use for pre-test genetic counseling, three of which also used the chatbot for risk assessment.[62] The remaining 3 of 7 studies only utilized the chatbot for risk assessment. The pooled completion rate for genetic cancer risk assessment across the 6 studies was 36.7%. Only one of the 4 studies with pre-test counseling specified that post-test genetic counseling was provided by a clinician.[43] For the remaining 3 studies, clinician involvement was not described, and mandatory counseling was not required to proceed with genetic testing.[64,67,68]

Several studies have evaluated post-test genetic counseling through chatbots. In Norway, a pre-test and post-test counseling chatbot was offered to individuals at risk for HBOC.[69] Almost all participants felt the chatbot was a helpful and empowering supplement to genetic counseling, viewing it as a particularly helpful alternative to independent internet searches. However, several participants noted that the lack of a "human touch" was a key limitation of the chatbot. Since the publication of these findings, two other studies have evaluated chatbots for post-test counseling. In Germany, a mobile app-based chatbot was offered to patients identified with HBOC as an adjunct to in-person genetic counseling.[70] The chatbot was designed to provide additional support following the in-person counseling session, with information topics personalized to the user's information, such as risk for males, preventative options, and fertility preservation. Acceptability of the chatbot was high, with particular appreciation for the chatbot's personalized conversations and the clear citation of references for all information provided. In the United States, Coen et al[71] developed a chatbot to facilitate post-test counseling for patients with positive screening results in South Carolina "In Our DNA SC" initiative, which has provided free genetic screening to over 100,000 individuals. The chatbot has not yet been tested in a patient population, but expert review of the tool demonstrated an average score of 3.86 on a 5-point Likert Scale across 8 predefined domains, with the lowest score on program accuracy and highest score on tone and usability. This study identified the need to improve the factual accuracy of information provided about In Our DNA SC's genetic testing program, before rollout for patients and integration into population-wide genomic screening.

In addition, one study explored the feasibility of ChatGPT to provide accurate genetic counseling in the gynecologic oncology context.[72] A list of 40 questions regarding genetic testing and counseling in the gynecologic oncology setting was curated in consultation with gynecologic oncologists and professional society websites. Questions ranged from general genetic counseling to queries specific to syndromes such as Lynch syndrome or HBOC. Responses were scored by 2 experts based on a scale of comprehensibility and accuracy, and compared with a final third reviewer to assign a score. ChatGPT was found to provide comprehensive and correct responses for 100% of the general counseling questions, but scored worse for HBOC-specific (64.7%) and Lynch-specific (66.6%) questions. These findings represent the potential utility of chatbots in genetic counseling for the gynecologic oncology context but highlight limitations in syndrome-specific genetic testing. Furthermore, the study did not evaluate the ability of AI-powered tools to provide accurate, acceptable genetic counseling at the individual level. Thus, to date, no studies have evaluated the ability of AI to provide analogous counseling to genetic counselors, accounting for an individual's specific testing results and clinical context.

CHALLENGES AND LIMITATIONS

Despite the promise of AI, several key challenges remain for the integration of AI-powered tools in genetic cancer risk assessment in gynecologic oncology. One of the primary limitations is ensuring the quality, completeness, and generalizability of algorithms.[39] The robustness of AI models depends on having sufficiently large and representative data sets for gynecologic cancers. The fragmentation of EMRs across different institutions, with data access restrictions and varying data collection protocols, impedes

the development of sufficiently large, heterogeneous databases.[73] Furthermore, many models are trained on cohorts of predominantly European ancestry based in high-resource health care settings, which limits generalizability to underrepresented populations.[39,40] This algorithmic bias can fail to capture variant pathogenicity and risk associations across diverse ancestry, potentially perpetuating disparities in genetic risk prediction, diagnostic accuracy, and health care outcomes.[33,74]

In addition, AI poses numerous ethical concerns around data privacy and usage. Genetic and genomic data are particularly sensitive, with information relevant to not only patients but also their biological relatives. Regulatory bodies must carefully develop governance frameworks around consent, data sharing, and reidentification risk, particularly when considering cross-institutional collaborations.[56,75] Strict data encryption and storage protocols are essential for ensuring data privacy and confidentiality.[76] The use of AI to guide risk assessment also raises the question of liability: if an AI system misclassifies risk, is responsibility placed upon the clinician, AI vendor, or institution?[77] Clear delineation of responsibilities and roles between chatbots and health care providers is required, with regular algorithmic audits and updates of models to ensure accuracy as new data become available.

FUTURE DIRECTIONS

Concerted efforts are required to expand data sets to include more ethnically diverse populations and to standardize data capture and annotation across clinical databases.[78] Although AI-powered chatbots hold potential to mitigate disparities in uptake of genetic services, these tools must become accessible and acceptable for vulnerable populations.[79] For example, this requires the development and testing of chatbots in languages other than English.[43] In addition, engagement with community advisory boards for underserved populations may help identify ways to tackle barriers towards uptake of chatbots, such as mistrust and limited engagement with the medical system.[80] To address the fragmentation of EMRs, federated learning offers an ML technique to collaboratively train models across decentralized servers while preserving data privacy.[75,77] In federated learning, models are trained locally (ie, an individual hospital system would generate a predictive model using its own patient data set). Each institution in the collaboration then shares the learned pattern—not the raw data—and these encrypted models are aggregated into a new set of parameters (a global model) that are then incorporated into the next round of training on local data sets.[75,81] Thus, raw data are not transferred across collaborating institutions, maintaining data privacy. In addition to federated learning models, homomorphic encryption allows AI models to compute encrypted data, preserving privacy across the computational workflow.[82]

However, technical issues such as nonindependent data and heterogeneity in data collection standards persist, and routine bias auditing remains critical to detect performance disparities across subgroups.[33,83] Further research on robust federated learning methods for genomic data is required, while standardized data schemas and metadata harmonization can support scalable model development.[77,83] To safely integrate chatbots into clinical practice, collaboration between clinicians, regulatory bodies, and AI technology developers is required. Together, these stakeholders must generate the legal and technical frameworks to address the aforementioned ethical and legal challenges.[75]

In addition, to incorporate AI tools into clinical practice, improving explainability and workflow logistics are both essential. Currently, deep learning models can remain a "black box" for clinicians, especially as model performance is evaluated by metrics such as AUCs, at the expense of interpretability.[40] To engender clinician and patient trust in AI-generated predictions, greater transparency is required. Explainable AI (XAI) aims to address this barrier.[33,84] XAI techniques identify critical features that influence model predictions such as patient demographics, specific genomic alterations, or inheritance patterns, which can in turn empower clinicians to validate AI-based predictions.[84] In parallel with these technical approaches, dialog-based explanations that facilitate clinician and patient decision-making are key.[85] For example, in addition to a pathogenicity score, annotation that explains why a VUS has been reclassified as likely pathogenic would provide more reassurance to clinicians and patients. Embedding AI within genetic counseling workflows and EMRs—where AI augments rather than replaces human counselors—can support scalability, equity, patient education, and shared decision-making.

Finally, the future of genetic cancer risk assessment lies in integrating not only germline genetic data but also epigenomic, transcriptomic, proteomic, radio-omic, pathologic, environmental exposure, and other EMR-derived data.[33] AI-driven multiomic integration will allow for more precise, dynamic risk prediction for gynecologic cancer that captures both inherited and acquired risk factors. Models operating on longitudinal data may also map risk trajectories and offer personalized preventive strategies over the life course.[86] To actualize this potential, collaboration is required between genomics, bioinformatics, data science, and clinical teams.

CONCLUSIONS

Artificial intelligence is rapidly transforming the landscape of genetic cancer risk assessment across oncologic fields, including gynecologic oncology. It offers unprecedented capabilities to integrate genomic, clinical, and population-level data. From automating genetic risk prediction from family histories to interpreting VUS to pre-test and post-test counseling, AI-driven tools have demonstrated clear potential to enhance the precision, efficiency, and equity of hereditary cancer care. However, actualizing the promise of this powerful tool requires overcoming key challenges such as data bias, interpretability, and ethical constraints that currently limit full clinical integration. Emerging frameworks such as federated learning and XAI offer the potential to address these barriers towards ensuring that AI-powered systems are not only scientifically rigorous but also socially responsible. As gynecologic oncology moves towards a future of AI-augmented precision medicine, interdisciplinary collaboration among clinicians, geneticists, data scientists, and ethicists will be essential to translate the potential of AI into real-world patient benefits.

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