AI and ML-Augmented Analysis For Precision Oncology and Cancer Diagnostics

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ABSTRACT

Each year, nearly 20 million people are diagnosed with a form of cancer, and this number is expected to significantly increase. Despite hundreds of years of research on various cancers, the causes and genetic influences behind them are still not fully understood and current treatment options are, at times, unsuccessful. The recent development of artificial intelligence (AI) and machine learning (ML) tools has the potential to accelerate knowledge of and advancements in cancer screening, diagnostics, and treatment. AI can be incorporated into complex sequencing data sets and clinical imaging to allow for earlier detection of cancer. Similarly, by applying these tools to clinical data, AI and ML can drastically improve diagnostics, allowing for more accuracy in identifying cancer types. Additionally, AI and ML can greatly improve patients' prognoses by utilizing deep learning to predict patient response to different treatments. In this review, I will discuss the integration of AI and ML technologies into cancer screening, diagnostics, and therapeutic approaches to enhance precision medicine and treatment outcomes for cancer patients.

INTRODUCTION

Cancer is one of the leading causes of death worldwide with nearly 20 million new cases arising each year (National Cancer Institute, 2025). In the United States alone, approximately 2 million people were diagnosed with a type of cancer in 2024 (Collins, 2024). Of these, 600,000 are anticipated to die from the cancer itself or related complications. Despite billions of dollars in funding, progress in developing new ways to combat cancer has been slow. The lack of significant advancements underscores the need for new approaches and perspectives in cancer research. Traditional approaches, including surgery, chemotherapy, and radiation therapy, are often non-specific and maladapted to the high degree of variability observed amongst cancers. More effective, precise, and personalized approaches could lead to better patient outcomes and decreased rates of death.

Artificial intelligence (AI) and Machine Learning (ML) have emerged as powerful tools in healthcare, offering the potential to process and analyze massive volumes of data generated through clinical care, research studies, and patient monitoring. AI refers to the general ability of computer systems to mimic human intelligence, such as performing complex tasks, reasoning, pattern recognition, and decision-making (Columbia Engineering, 2025). ML, which is a subset of AI, involves the use of algorithms that learn insights and recognize patterns from data in order to maximize the performance of tasks. The use of ML allows machines to be trained to autonomously learn from past data with the goal of delivering accurate results. While ML relies on statistical models and has a more limited scope of applications, both AI and ML offer powerful benefits of integrating heterogeneous data and informing decision-making.

In healthcare, they are used to analyze complex information, such as genomic data and electronic health records (EHRs); identify patterns; and support faster, more accurate diagnoses and treatments. AI and ML have the potential to revolutionize cancer care by uncovering previously unknown oncogenes and tailoring care to individuals depending on their cancer profiles, a strategy known as precision oncology. This could significantly improve diagnostic accuracy and reduce mortality. As AI and ML continue to expand and improve, it is critical to assess their impact on cancer care. In this review, I will examine the role of AI and ML in precision oncology, focusing specifically on their ability to improve cancer detection, diagnostic accuracy, and personalized treatment strategies.

MAIN TEXT

Revolutionizing Cancer Screening with AI and ML Technologies

Early cancer detection can improve survival rates, but traditional screening methods often miss subtle signs. Advanced tools such as AI and ML can enhance accuracy and efficiency of detecting these signs. Many new sequencing techniques, such as ARTEMIS and EPIC-seq, are being developed that allow for better detection of cancer DNA and the integration of AI and ML technologies.

DNA Analysis Techniques

ARTEMIS, which stands for Analysis of RepeaT EleMents in dISease, analyzes the parts of DNA that were once thought to have no function. These regions of non-coding DNA have been discovered to have specific functions and play a role in tumor cells. Parts of non-coding DNA have repetitive sequences, known as repeat elements, located throughout the genome, which have been found to be altered in cancerous tissue. ARTEMIS was designed to specifically identify these repeat elements, in addition to circulating cell-free DNA (cfDNA) - fragments of DNA shed from tumors that float in the bloodstream - in order to find tumor-specific changes in cancer patients (Annapragada et al., 2024). The ARTEMIS ML model was used to analyze over 1,200 types of repeat elements comprising nearly half of the entire human genome. It identified a large number of repeats that were altered in tumor formation (Johns Hopkins Medicine, 2024). When tested on patients with various cancers, ARTEMIS, when combined with another DNA fragment assay, was able to predict patients' tumor types with 75-85% accuracy. This technology allows researchers to analyze the entire genomes of patients in a fast and efficient manner, allowing for a noninvasive method to detect cancer.

Another sequencing method called epigenetic expression inference from cell-free DNA-sequencing (EPIC-seq) was developed to detect cfDNA in the bloodstream. Similar to ARTEMIS, EPIC-seq aims to be a noninvasive method for cancer detection. Its ability to classify different tumor subtypes from just blood samples shows EPIC-seq's ability as a powerful tool in early, non-invasive cancer screening. This tool can be used to analyze biological markers faster, and more precisely, than traditional methods. EPIC-seq relies on sequencing the promoter regions of genes of interest of the cfDNA to predict differential gene expression in tumors (Esfahani et al., 2022). The sequencing results are then combined with machine learning to predict RNA expression and identify the tumor subtype. This method demonstrated high performance within cancer detection and tumor classification, with area under the curve (AUC) values above 0.9 for multiple cancer types. While EPIC-seq utilizes cell-free DNA, other AI models use other biological markers to detect tumors.

Epigenetic Targeting

DNA methylation represents a promising biomarker for revolutionizing cancer screening through AI and ML technologies, as tumors exhibit distinct methylation patterns that can be detected using computational approaches. While traditional methylation analysis has shown promise - such as a cervical cancer study that identified three methylation markers (GHSR, SST, and ZIC1) with ROC curve values between 0.86 and 0.89 for detecting precancerous lesions (Verlaat et al., 2017) - the integration of artificial intelligence has dramatically enhanced detection capabilities. A study demonstrated this potential by applying different AI algorithms, including support vector machines and deep learning, to DNA methylation patterns in circulating cell-free DNA from lung cancer patients (Bahado-Singh et al., 2022). These AI-driven approaches achieved exceptional performance with area under the curve (AUC) values of 1.00 and 100% sensitivity and specificity, representing a significant advancement over traditional screening methods. This integration of AI with epigenetic biomarkers exemplifies how machine learning technologies are transforming cancer screening by enabling highly accurate and minimally invasive detection methods that could revolutionize early cancer diagnosis.

Based on recent comprehensive research by Dr. Hajjar and colleagues, artificial intelligence and machine learning technologies are fundamentally revolutionizing cancer screening through multi-cancer early detection (MCED) tests that overcome traditional diagnostic limitations (Hajjar et al., 2024). These AI-augmented liquid biopsy platforms leverage sophisticated algorithms - including ensemble methods and deep learning - to analyze complex biomarker signatures from blood and urine samples, enabling simultaneous detection of multiple cancer types from a single test. Machine learning approaches have demonstrated remarkable performance with sensitivity ranges of 84-100% for early-stage cancers and specificity rates exceeding 98%, while AI-driven methylation pattern analysis has achieved clinical validation through FDA-approved tests like Galleri. Notably, multi-modal approaches integrating diverse biomarker types, such as the combination of protein markers with circulating tumor DNA and epidemiological data, have yielded superior diagnostic accuracy compared to single-biomarker strategies. This transformation from invasive, single-cancer screening to minimally invasive, AI-powered multi-cancer detection represents a paradigm shift toward precision oncology, offering the potential to dramatically improve early detection rates and save lives through quick intervention.

The integration of artificial intelligence and machine learning technologies has transformed cancer screening methodologies, offering unprecedented capabilities in early detection through advanced genomic analysis. AI algorithms excel at processing the massive datasets generated by next-generation sequencing, enabling more accurate and efficient identification of cancer-causing variants through improved variant-calling techniques such as DeepVariant, which outperforms standard tools in detecting significant genetic alterations (Lin et al., 2022).

Revolutionary screening approaches now leverage liquid biopsy technology, where AI facilitates the analysis of circulating tumor cells, extracellular vesicles, and tumor-derived cell-free DNA (cfDNA) found in blood and other bodily fluids, providing a less invasive alternative to traditional tissue sampling (Ginghina et al., 2022). Multi-cancer early detection (MCED) tests utilize AI to analyze methylation patterns indicative of cancer in cfDNA from asymptomatic populations, with machine learning algorithms processing the vast amounts of data to enable timely analysis and intervention (O'Connor & McVeigh, 2025). Advanced AI models like AlphaMissense can predict the pathogenicity of all possible missense variants in the human genome at a single amino acid substitution level, allowing clinicians to identify and screen for clinically relevant pathogenic variants with unprecedented precision (Cheng et al., 2023). Computer vision applications further enhance screening capabilities by analyzing histopathological and radiological images to predict genetic mutations within tumors, as demonstrated by models that can determine EGFR mutation status in non-small-cell lung cancer from CT and PET scan images (Popat et al., 2025). These AI-driven screening innovations collectively enable earlier cancer detection, more comprehensive genomic profiling, and the potential for population-wide screening programs that could greatly improve patient outcomes through timely intervention.

AI and ML-Powered Advances in Cancer Diagnostics

AI and ML are transforming diagnostics by making detection faster, more accurate, and more personalized. These technologies can analyze vast amounts of complex data, including medical scans, genetic profiles, and pathology slides, to uncover patterns that may be difficult for human experts to detect. Recent advances have demonstrated how AI can improve diagnostic precision, classify cancer subtypes more effectively, and predict treatment outcomes. As these tools continue to evolve, growing attention is being placed on how AI and ML models, such as Explainable AI, can help patients receive more efficient diagnoses.

The Use of Explainable AI

Explainable AI (XAI) helps patients receive faster diagnoses by making complex machine learning predictions more transparent and actionable for clinicians. A comparison of multiple XAI platforms used on breast cancer datasets revealed one superior AI model, SHapley Additive exPlanations (SHAP), that was able to clearly present model outputs in simpler terms for clinicians to understand (Ghasemi et al., 2024). By showing how specific features, such as biomarkers or clinical variables, contribute to a diagnosis, SHAP allows healthcare providers to interpret AI-generated results while also having a clearer understanding of the process that the AI model used to generate those outputs. This interpretability builds trust in the model's findings, enabling doctors to make faster and more confident decisions. As a result, patients can move more quickly from initial screening to diagnosis and treatment, reducing delays that could potentially worsen patients' outcomes. In time-sensitive conditions like breast cancer, this acceleration in the diagnosis process can make a critical difference in prognosis, highlighting how XAI not only improves model transparency but also has a direct impact on patient care. In addition to faster diagnoses, AI is also being used to anticipate complications during treatment, helping to predict negative side effects that can result from these life-saving therapies.

Targeting Specific Cancer Types

AI is also being used to support treatment continuity by predicting and preventing complications that might otherwise delay or interrupt therapy. In a phase 3 randomized controlled trial, researchers evaluated the use of romiplostim (ROMI), a thrombopoietin receptor agonist, to address chemotherapy-induced thrombocytopenia (CIT) in patients with gastrointestinal cancers. CIT is a common and serious side effect of chemotherapy that can lead to dose reductions, delays, or even discontinuation of treatment, potentially compromising patient outcomes. The study found that patients receiving ROMI were significantly more likely to avoid CIT-related chemotherapy modifications compared to those receiving placebo - 84% versus 36%, respectively (Al-Samkari, 2025). This kind of clinical challenge is increasingly being addressed with AI models that can anticipate risks, like CIT, based on patient data. When integrated into care, such predictive tools can help providers make early decisions, such as adjusting therapy or preemptively introducing supportive treatments like ROMI, to keep patients on track. This demonstrates the growing role of AI in optimizing treatment strategies, a trend also reflected in recent breakthroughs in personalizing therapies for many other cancer types.

Recent advances show that AI can also play a critical role in identifying which patients are most likely to benefit from specific cancer therapies, helping to personalize treatment while avoiding unnecessary side effects. In a large international study, researchers developed an AI tool that analyzes tumor biopsy images to predict which patients will respond best to abiraterone - a widely used drug for treating prostate cancer. Although abiraterone has already improved survival for many with advanced disease, it also comes with risks such as high blood pressure, liver abnormalities, and increased chances of heart problems (Gregory, 2025). The AI test identifies a subgroup of patients - about 25% - who are most likely to benefit from treatment, with data showing the drug cut their five-year mortality risk nearly in half (Gregory, 2025). Meanwhile, for patients unlikely to benefit, the tool helps avoid overtreatment by showing that standard therapies alone are sufficient. This approach reflects a broader shift toward AI-powered precision medicine, where treatment decisions are guided not just by clinical guidelines but by individualized predictions derived from routinely collected data. By enabling clinicians to match therapies more effectively to each patient's specific disease profile, AI helps improve outcomes, reduce toxicity, and make better use of healthcare resources.

Pancreatic cancer - particularly pancreatic ductal adenocarcinoma (PDAC) - remains one of the most aggressive and lethal forms of cancer, with a five-year survival rate of just 13% (American Cancer Society, 2025). Diagnosis is especially difficult due to vague early symptoms and a lack of reliable screening methods, resulting in 80-90% of patients being diagnosed at an advanced stage when curative treatment is no longer an option. Although CT imaging is the most commonly used modality for initial evaluation and offers a sensitivity of 70-90%, its diagnostic effectiveness is limited without advanced analytical support (Chen et al., 2016). To address these challenges, one study presents a deep learning-based diagnostic system that integrates Convolutional Neural Networks (CNN) with a YOLO-based CNN architecture (YCNN) to enhance early detection of PDAC (Dinesh et al., 2023). By analyzing CT scans and identifying cancerous features using pixel-level classification, the system demonstrated superior performance, achieving F1-scores of 0.99 and 1.00 in detecting and grading pancreatic cancer. The model processes images by dividing them into 224×224 pixel patches, classifying them, and then reconstructing the complete image for review by pathologists, offering both precision and interpretability. The use of platforms like PyCharm and Google Colab streamlines development and accessibility, and the authors propose future enhancements including data augmentation, expanded datasets, and synthetic image generation guided by specialists. Ultimately, this AI-powered system holds promise as a practical diagnostic aid, capable of providing consistent second opinions through a user-friendly web interface and improving the early detection and classification of pancreatic cancer.

The Use of Omics Data

Machine learning is now underpinning major advances in cancer diagnostics by enabling integration of heterogeneous omics data - genomic, transcriptomic, proteomic, and more - into unified analytical frameworks. According to Dr. Cai and colleagues, these approaches are generally grouped into general-purpose methods, which combine dimensionality reduction with downstream modeling, and task-specific models, which are end-to-end tools tailored to particular applications (Cai et al., 2022). Central to this integration is the strategy known as middle integration, which avoids naive feature concatenation while consolidating signals across omics layers through machine learning, mitigating the curse of dimensionality common in multi-omics datasets. The review also benchmarks five tools, including DIABLO, MOFA2, iClusterPlus/Bayes, and moCluster, used on data from the Cancer Cell Line Encyclopedia, evaluating for their accuracy in cancer type classification and drug response prediction, as well as runtime performance. Notably, DIABLO, a supervised method that leverages label information, like tissue type, achieved the highest cancer-type prediction accuracy (78.7%), underscoring how integrative ML models can more precisely classify tumor subtypes compared to conventional unsupervised approaches (Cai et al., 2022). These developments illustrate how AI-driven integration of multi-omics datasets delivers richer molecular diagnostics, enabling identification of distinct cancer subtypes, more accurate treatment stratification, and ultimately a path toward truly personalized oncology.

AI and ML Applications in Tailored Cancer Therapies

Artificial intelligence and machine learning are playing an increasingly important role in developing and refining cancer treatment strategies. By processing large-scale data from clinical records, genetic information, and histopathological images, AI/ML models can identify complex patterns that predict how individual patients will respond to specific therapies. This allows for more personalized and effective treatment plans, going beyond generalized protocols. From enhancing chemotherapy response predictions to identifying biomarkers for immunotherapy, AI and ML are accelerating the shift toward precision oncology.

Predicting Patient Response to Treatment

One example of this progress is a new AI tool that predicts which cancer patients are most likely to benefit from immune checkpoint inhibitors, which are a type of immunotherapy drug, using only routinely collected clinical data (Chang et al., 2024). These therapies help the immune system recognize and attack cancer cells, but their effectiveness varies widely between individuals. Existing tests to predict treatment response are often expensive or unreliable. To address this, the team created a ML-based scoring system called LORIS (LOgistic Regression-based Immunotherapy-response Score), which uses six routinely collected clinical features: tumor mutational burden, patient age, cancer type, history of prior therapies, blood albumin levels, and blood NLR (a measure of inflammation). After evaluating data from over 2,800 patients with 18 different cancer types, LORIS outperformed existing models in predicting both short- and long-term outcomes. Its simplicity and accessibility make it a promising tool for guiding personalized treatment decisions in everyday clinical practice. Beyond tools like LORIS, broader applications of AI are also advancing immunotherapy by identifying predictive biomarkers and new therapeutic targets.

A similar machine learning system has been built by Dr. Chowell's team out of the Icahn School of Medicine at Mount Sinai called SCORPIO (Standard Clinical and labOratory featuRes for Prognostication of Immunotherapy Outcomes). SCORPIO predicts how well cancer patients will respond to immune checkpoint inhibitors using only basic, routinely collected clinical data, such as blood counts and metabolic profiles (Yoo et al., 2025). Unlike traditional biomarkers such as evaluating tumor mutational burden and PD-L1 expression - which are expensive, time-consuming, and not widely available methods - SCORPIO uses accessible data and outperforms these methods in predicting patient response. Validated on nearly 10,000 patients across 21 cancer types, SCORPIO represents a scalable and cost-effective tool that could expand access to precision oncology, especially in limited-resource conditions. Additionally, SCORPIO has the capability to accurately predict survival rates of patients over the course of 2.5 years, with performance rates of about 75%. Researchers emphasize its potential to reduce unnecessary treatments, improve outcomes, and promote equitable cancer care by making personalized treatment decisions possible in daily clinical practice.

Artificial intelligence and machine learning are changing how we approach personalized cancer treatment by making it possible to tailor therapies with incredible precision. In cancer treatment, especially chemotherapy, AI helps analyze how medications interact with individual patients, making sure treatments are given effectively while predicting how well patients will tolerate and respond to them (Abdul Rasool Hassan et al., 2025). These AI tools excel at processing huge amounts of patient information – including genetic profiles, biomarker levels, and medical histories – to figure out the best treatment combinations and predict how each person will respond. Machine learning can spot complex patterns in tumors and patient biology that doctors are rarely able to catch on their own, leading to better drug options, optimized doses, and precise timing of treatments. Additionally, AI systems are continuously learning from treatment results across thousands of patients, allowing them to improve prediction accuracy, find alternative ways to target cancers, and assist with overcoming drug resistance. This smart approach to cancer care represents a major shift away from the old "same treatment for everyone" model to truly personalized medicine, where treatment decisions are made by sophisticated algorithms that consider both the unique fingerprint of each person's cancer and their individual medical background.

AI is also being used to support treatment continuity by predicting and preventing complications that might otherwise delay or interrupt therapy. In a phase 3 randomized controlled trial, researchers evaluated the use of romiplostim (ROMI), a thrombopoietin receptor agonist, to address chemotherapy-induced thrombocytopenia (CIT) in patients with gastrointestinal cancers. CIT is a common and serious side effect of chemotherapy that can lead to dose reductions, delays, or even discontinuation of treatment, potentially compromising patient outcomes. The study found that patients receiving ROMI were significantly more likely to avoid CIT-related chemotherapy modifications compared to those receiving placebo - 84% versus 36%, respectively (Al-Samkari, 2025). This kind of clinical challenge is increasingly being addressed with AI models that can anticipate risks, like CIT, based on patient data. When integrated into care, such predictive tools can help providers make early decisions, such as adjusting therapy or preemptively introducing supportive treatments like ROMI, to keep patients on track. This demonstrates the growing role of AI in optimizing treatment strategies, a trend also reflected in recent breakthroughs in personalizing therapies for many other cancer types.

The Use of Deep Learning

A novel deep learning (DL) model demonstrates how artificial intelligence can significantly enhance treatment prediction for patients with metastatic clear cell renal cell carcinoma (ccRCC), the most common and lethal form of kidney cancer. ccRCC is typically treated with anti-angiogenic (AA) therapies, which aim to block the formation of new blood vessels feeding the tumor. However, due to substantial variability in tumor vascular architecture and gene expression, identifying which patients will benefit from AA therapy has remained a major clinical challenge. Traditional approaches, such as RNA-based tools like the Angioscore, provide some predictive power but are expensive, time-consuming, and sensitive to tumor sampling variability. To overcome these limitations, researchers developed the H&E DL Angioscore using a convolutional neural network trained on routine hematoxylin and eosin (H&E)-stained pathology slides (Jasti et al., 2025). This AI model automatically quantifies angiogenesis and generates a continuous score that predicts patient response to AA therapies. In both real-world and clinical trial cohorts - including the IMmotion150 trial - the model performed comparably to the RNA Angioscore (c-index 0.66 vs. 0.67) and outperformed conventional vascular biomarkers like CD31 immunohistochemistry (Jasti et al., 2025). Beyond its predictive accuracy, the model produces visually interpretable vascular heatmaps that highlight intra-tumoral variation, making its output more clinically transparent and trustworthy. By enabling fast, scalable, and cost-effective assessment of tumor angiogenesis, the H&E DL Angioscore represents a powerful step forward in personalizing treatment for renal cancer patients and optimizing the use of AA therapies in clinical practice.

Traditional cancer treatment strategies, such as those based on maximum tolerated dose (MTD), often fail in the metastatic setting due to rapid emergence of drug resistance. To address this limitation, researchers developed a deep reinforcement learning (DRL) framework designed to optimize adaptive therapy (AT), an approach that delays resistance by maintaining a population of drug-sensitive cancer cells to suppress resistant ones (Gallagher et al., 2024). Using a virtual cohort of prostate cancer patients, the study trained DRL agents on tumor dynamics simulated by mechanistic mathematical models. These agents learned personalized treatment schedules that significantly outperformed both MTD and commonly used AT strategies, including outperforming AT50 in delaying disease progression. Notably, the DRL policies required only a single input – the tumor burden threshold – and were interpretable, allowing clinicians to understand and trust the model's decisions. Even when trained without full knowledge of the tumor model, the DRL framework achieved robust and clinically relevant outcomes, highlighting its flexibility. To support clinical translation, the authors proposed a five-step pathway emphasizing mechanistic modeling, DRL training, and clinical interpretability. This work shows how AI-guided strategies can transform cancer care by adapting therapy to the evolving dynamics of each individual patient's disease. Together with earlier advances in drug response prediction and treatment stratification, these innovations demonstrate that AI is reshaping cancer therapy into a more dynamic, data-driven process. As machine learning models grow more interpretable and clinically validated, they are poised to become essential tools in designing effective, patient-specific treatment plans.

CONCLUSION

The growing integration of AI and ML into cancer research and care is reshaping how clinicians detect, diagnose, and treat malignancies. In screening, these technologies have advanced the analysis of genomic, epigenetic, and molecular data, enabling earlier identification of cancer-related patterns that may be missed by traditional approaches. In diagnostics, AI has improved the accuracy of classification, integrated diverse biological datasets, and provided explainable outputs that enhance trust in clinical settings. In treatment, AI models, including deep learning, have optimized therapy schedules, predicted patient-specific drug responses, and tailored interventions to tumor dynamics. Together, these developments mark a decisive move toward precision oncology, where care is increasingly proactive, data-driven, and individualized.

The advantages of AI and ML in oncology are substantial. They allow rapid processing of high-dimensional data, reveal patterns invisible to human interpretation, and offer the potential for more accurate and timely decision-making. Personalized approaches informed by AI can reduce overtreatment, improve survival outcomes, and enhance quality of life. However, limitations remain. Many AI models are developed on controlled datasets that may not reflect the complexity of real-world patient populations. Data privacy concerns, potential biases in training datasets, and limited interpretability in some algorithms can hinder clinical adoption. Additionally, the need for large, high-quality annotated datasets presents logistical and ethical challenges that must be addressed.

To make these technologies mainstream, several steps are essential. Rigorous clinical validation across diverse populations is needed to confirm the reliability and generalizability of AI models. Interdisciplinary collaboration between clinicians, data scientists, and regulatory bodies will be important to ensure that algorithms meet both performance and ethical standards. Efforts to improve model transparency and interpretability will foster clinician and patient trust. Integrating AI tools into existing healthcare infrastructure, supported by data governance frameworks, will further smooth the transition from research to clinical practice. As these requirements are met, AI and ML have the potential to become indispensable components of oncology, delivering real benefits to cancer patients worldwide.

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