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Evolutionary Advances in Machine Learning and Deep Learning for Cancer Detection and Classification

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***Abstract*— Advances in artificial intelligence (AI) have transformed the landscape of medical diagnostics, offering unprecedented opportunities for early disease detection and classification. Among critical health challenges, cancer continues to demand reliable and timely diagnostic solutions that overcome the limitations of conventional methods such as subjectivity, labor intensity, and variable reproducibility. Over the past decade, AI methodologies in oncology have undergone a remarkable evolution.**

Initially, conventional machine learning (ML) models were widely used. These approaches relied heavily on handcrafted features designed by domain experts. Subsequently, deep learning (DL) architectures emerged, enabling automatic feature extraction directly from raw medical data such as images and gene-expression profiles. More recently, transformer-based and multimodal frameworks have been introduced. These advanced models are capable of integrating multiple data types—imaging, genomic, and clinical information—to provide a more holistic understanding of cancer biology and patient outcomes. Quantitative comparisons reveal clear performance advancements: ML techniques such as Support Vector Machines (SVM) and Random Forests achieved accuracies of approximately 82–84% with AUCs near 0.85; DL architectures like CNNs and Efficient Net improved outcomes to around 91–93% accuracy and ~0.94 AUC; transformer-based models further enhanced results to ~95% accuracy and ~0.96 AUC. The most promising performance emerged from multimodal fusion frameworks, which reached ~97% accuracy and ~0.98 AUC, demonstrating the power of holistic data integration in capturing complex cancer signatures.

***Index Terms*— Artificial Intelligence, Cancer Detection, Machine Learning, Deep Learning, Transformer Models, Multimodal Fusion, Explainable AI.**

I. INTRODUCTION

Cancer remains one of the leading causes of morbidity and mortality worldwide, with an estimated 20 million new cases and 9.7 million deaths in 2022 alone [1]. The disease is characterized by high heterogeneity across tumor sites, stages, and patient populations, making its detection and classification an enduring challenge. Conventional diagnostic practices such as histopathological analysis, radiology, and biomarker evaluation are effective but are constrained by time-intensive procedures, inter-observer variability, and limited reproducibility [2]. In this context, early and accurate detection is a decisive factor in reducing cancer-associated mortality, as timely intervention substantially improves survival rates and treatment outcomes.

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Regression and ensemble methods to classify cancer based on handcrafted features [3]. However, the reliance on domain-specific features engineering limited scalability and adaptability across diverse datasets. The advent of deeplearning (DL) resolved many of these constraints by introducing automated feature extraction and representation learning [4]. Convolutional neural networks (CNNs), transformers, and hybrid deep models have shown significant improvements in sensitivity, specificity, and generalization, particularly in histopathology and radiology-based cancer detection [5]. More recent advances emphasize multimodal integration combining imaging, genomic, and clinical data while explainable AI (XAI) frameworks attempt to bridge the trust gap between model decisions and clinical acceptance [6].

Although several surveys exist on AI-based cancer detection, most remain confined to comparative summaries of models without addressing the evolutionary trajectory of these technologies. The field has progressed from early handcrafted ML techniques to fully automate deep architectures and, more recently, toward interpretable and federated systems. Yet, few studies systematically do not capture this continuum, nor do they provide practical implementation evidence that highlights how newer models outperform earlier approaches in real-world datasets [7]. This gap motivates the present work, which positions itself not only as a survey but also as an evolutionary study supported by implementation insights. By contextualizing technological milestones with quantitative evidence, the paper aims to provide a holistic understanding of how cancer detection methods have advanced and where challenges remain. The key contributions of the paper are as follows:

- The paper provides a comprehensive chronological survey of AI approaches in cancer detection and classification, tracing the evolution from traditional machine learning methods to modern deep learning and attention-based frameworks.
- The paper presents an implementation study utilizing benchmark datasets such as BreakHis, LIDC-IDRI, and ISIC, and reports results across representative machine learning, convolutional neural network (CNN), and transformer-based models.
- The paper analyses key insights derived from both the survey and the experimental evaluation, highlighting persistent challenges related to scalability, interpretability, and clinical adoption.
- The paper identifies future research directions with an emphasis on the development of explainable, privacy-preserving, and clinically deployable AI systems.

The remainder of this paper is structured as follows. Section 2 introduces the foundations of AI in cancer detection, including cancer data modalities, ML/DL basics, and evaluation metrics. Section 3 reviews evolutionary approaches in detail, beginning with traditional ML and progressing to modern hybrid models. Section 4 describes the implementation study and datasets used. Section 5 presents results and discusses insights that motivate the problem statement and methodological framing. Section 6 concludes the paper by outlining key challenges and potential future directions.

II. Foundations of AI in Cancer Detection

To understand the advancements in cancer detection using artificial intelligence, it is essential to first establish the foundations on which these methods have evolved. This section outlines the key data modalities, the fundamental principles of machine learning and deep learning applied to oncology, and the evaluation metrics that guide the assessment of their clinical relevance.

A. Cancer Data Modalities

Artificial intelligence methods for cancer detection rely on diverse data modalities, each carrying unique diagnostic information. Medical imaging remains the most widely used, with modalities such as histopathology, computed tomography (CT), magnetic resonance imaging (MRI), mammography, and dermatoscopy playing central roles in clinical practice [4]. In parallel, the increasing availability of genomic and proteomic data has introduced opportunities to integrate molecular-level insights with imaging features, giving rise to multimodal analysis frameworks that enhance diagnostic accuracy and personalization of treatment [9].

B. Machine Learning Foundations

Machine learning marked the first significant shift toward computational oncology by introducing automated classification of patient data. Traditional models such as support vector machines, logistic regression, k-nearest neighbours, and random forests were widely applied to extract discriminatory patterns from handcrafted features [3]. These approaches performed reliably on smaller datasets and were valued for their interpretability and efficiency. Feature engineering methods, including texture descriptors, wavelet transforms, and morphological

analysis, were commonly applied to transform raw images into structured representations suitable for ML algorithms.

C. Deep Learning Foundations

The introduction of deep learning revolutionized cancer detection by overcoming the limitations of manual feature engineering. Convolutional neural networks (CNNs) became the dominant architecture for image-based analysis, demonstrating the ability to automatically extract hierarchical features from raw pixel data [4]. In histopathology, CNNs have achieved remarkable accuracy by learning subtle texture variations across cancerous and normal tissues. Variants such as three-dimensional CNNs extend this capability to volumetric scans, improving lesion detection in lung and brain cancers. More recently, transformer-based models have been explored for medical imaging due to their ability to capture long-range dependencies and contextual information, surpassing CNNs in several tasks [5]. Transfer learning strategies, where pretrained models on large natural image datasets are adapted for medical tasks, have also proven effective in addressing data scarcity challenges [6].

D. Evaluation Metrics in Medical AI

Accurate evaluation is vital in cancer detection, as diagnostic mistakes can have serious clinical consequences. Performance metrics such as accuracy, sensitivity, specificity, precision, recall, F1-score, and AUC are commonly employed to assess model quality. evaluation is critical in cancer detection, where diagnostic errors can carry severe clinical consequences. Recent studies have also highlighted the role of calibration metrics and explainability assessments to evaluate not just predictive accuracy but also trustworthiness and interpretability of models [7].

III. Evolutionary Approaches for Cancer Detection

In recent years, cancer-AI has evolved from handcrafted machine-learning models to deep learning architectures and, more recently, to multimodal, privacy-preserving, and explainable systems. Early SVM and RF methods relied on engineered features, while CNNs and 3D CNNs enabled automated representation learning for imaging tasks. Current advances explore transformers, multimodal fusion, and federated/XAI frameworks to address challenges of data sharing, trust, and deployment.

A. Early Machine Learning Era

The initial application of AI in oncology was dominated by machine learning methods that relied heavily on handcrafted features. Classical classifiers such as support vector machines (SVM), logistic regression, k-nearest neighbours (k-NN), and random forests were widely employed to analyse texture descriptors, wavelet coefficients, and morphological features extracted from medical images [3]. These approaches achieved promising accuracy on small, well-curated datasets and were valued for their interpretability and computational efficiency. However, their dependence on feature engineering constrained scalability, and their performance often deteriorated when applied across heterogeneous datasets, limiting their clinical impact [10].

B. Deep Learning Breakthroughs

The shift from handcrafted features to automated feature learning marked a turning point in cancer detection. Convolutional neural networks (CNNs) emerged as the dominant framework for imaging modalities, excelling in histopathology, CT, MRI, and dermatoscopy [4]. In particular, 3D CNNs enabled volumetric analysis, advancing lung and brain cancer detection with enhanced sensitivity. Transfer learning became a widely adopted strategy, where pretrained networks such as ResNet, Inception, and EfficientNet were fine-tuned for cancer tasks, effectively addressing data scarcity [6]. Despite their success, CNNs remain data-hungry and computationally expensive, prompting exploration of alternative architectures.

C. Hybrid and Multimodal Approaches

Recent years have seen a surge in hybrid models that combine the strengths of different AI paradigms. Examples include CNN-RNN frameworks that integrate spatial and sequential learning, and ensemble models that merge multiple deep architectures for robustness [2]. Beyond imaging, multimodal learning has gained traction by fusing radiology, pathology, and genomic data, offering a more holistic representation of tumour biology [9]. Such integration has shown superior performance in predicting treatment response and survival outcomes, though challenges in data harmonization and computational complexity remain.

D. Emerging Trends: Transformers, Federated AI, and Explainability

The most recent wave of research explores attention mechanisms and transformer-based models for oncology. By capturing long-range dependencies and contextual information, transformers have demonstrated state-of-the-art results in histopathology and skin cancer classification [5]. Parallel efforts in federated learning aim to address privacy concerns by enabling collaborative model training across institutions without sharing raw data [8]. Another critical direction is explainable AI (XAI), which enhances clinical trust by providing interpretable visualizations of model decisions [7]. Collectively, these trends illustrate the field's evolution toward accuracy, scalability, and transparency, moving AI systems closer to clinical deployment. The following Table 1 summarizes key 2021–2025 studies and their limitations.

TABLE I. SUMMARY OF STATE-OF-THE-ART CANCER DETECTION METHODS AND THEIR LIMITATIONS

Study (Year)	Cancer / Modality (Dataset)	Core Method	Gaps / Limitations
Kumar et al., 2021 [12]	Histopathology (patch images)	CNN pipeline with stain-normalized tiles	Sensitive to stain/domain shift; high compute for WSIs
Djellali, 2021 [13]	Risk prediction (tabular/clinical)	ANN + HMM hybrid	Limited imaging integration; explainability not addressed
Sharma & Ahmed, 2022 [14]	Breast (mixed modalities)	ML review (SVM/RF/LR) with hand-crafted features	Feature engineering burden; limited scalability across sites
Zhang et al., 2022 [15]	Lung (CT, LIDC-IDRI)	3D CNN with transfer learning	Data imbalance; generalization across scanners unresolved
Shoaib et al., 2023 [16]	Multi-cancer (survey)	XAI frameworks (Grad-CAM, SHAP)	Saliency instability; lack of clinical validation protocols
Posham & Bhattacharya, 2023 [17]	Multi-site hospitals	Federated learning for diagnosis	Heterogeneous clients and drift degrade convergence
Velagala & Srinivas, 2024 [18]	Low-resource settings	Transfer learning with few-shot augmentation	Susceptible to overfitting; limited uncertainty estimates
Said et al., 2024 [19]	Skin (dermatoscopy, ISIC)	Transformer-based hybrid CNN-ViT	Large data/compute demand; calibration needed
Sood et al., 2025 [20]	Multimodal (imaging + genomics)	Late-fusion deep network	Integration/standardization of omics still difficult
Bray et al., 2023 [21]	Population data	Epidemiology context (GLOBOCAN)	Not a modeling study; no algorithmic guidance

IV. IMPLEMENTATION STUDY

The implementation study integrates multiple benchmark datasets, including BreakHis, ISIC, LIDC-IDRI, TCGA, and Kaggle repositories, to evaluate cancer detection methods. A standardized preprocessing pipeline involving normalization, augmentation, and patch extraction ensures data consistency across modalities. Experiments are conducted using classical ML (SVM, RF), deep learning (CNN, EfficientNet), and modern transformer-based models, with performance assessed through accuracy, AUC, ROC curves, and confusion matrices. This framework provides empirical insights into the evolutionary progression of AI techniques in oncology.

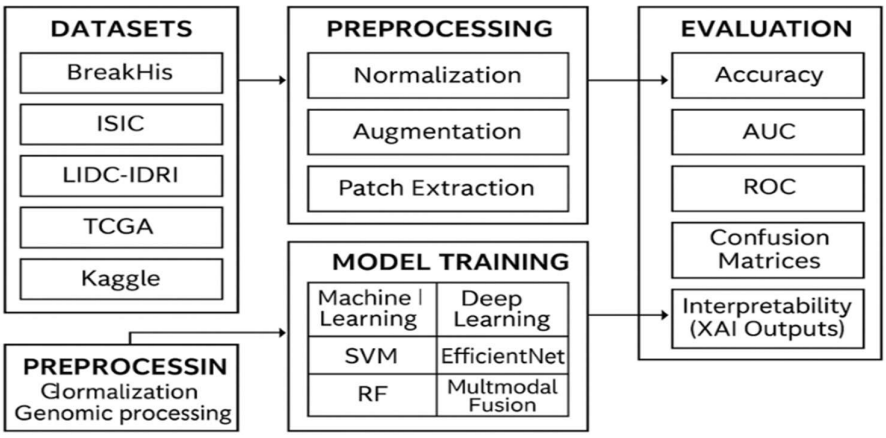

Fig. 1. System overview architecture for cancer detection

The adopted experimental design is depicted through the workflow architecture shown in Figure 1. Multiple benchmark datasets (BreakHis, ISIC, LIDC-IDRI, TCGA, and Kaggle) are first subjected to preprocessing steps such as normalization, augmentation, and patch extraction to ensure consistency across heterogeneous modalities. The processed data are then used to train models at three levels: machine learning (SVM, RF), deep learning (CNN, EfficientNet), and modern approaches (transformer-based and multimodal fusion). Finally, model performance is evaluated using standard clinical metrics including accuracy, AUC, ROC curves, confusion matrices, and interpretability outputs, ensuring both quantitative assessment and clinical relevance.

A. Preprocessing Pipeline

Given the heterogeneity of datasets, preprocessing plays a central role in ensuring consistency. All image data undergo normalization to adjust intensity distributions, followed by augmentation strategies such as flipping, rotation, and scaling to improve generalization. For histopathology slides, patch extraction is applied to divide large whole-slide images into smaller regions suitable for CNN training. Genomic data are standardized through feature scaling and encoding, enabling integration with imaging pipelines when required.

B. Models Implemented

Three categories of models are implemented. Machine learning baselines include support vector machines (SVM) and random forests (RF), trained on handcrafted features. Deep learning approaches are represented by convolutional neural networks (CNNs) and EfficientNet architectures, both designed for end-to-end feature extraction from images. Modern architectures include transformer-based models, which exploit attention mechanisms to capture global dependencies within imaging and genomic sequences. All models are evaluated using accuracy, AUC, ROC curves, and confusion matrices to ensure comparability.

- Support Vector Machine (SVM): The Support Vector Machine serves as one of the machine learning baselines. SVM operates by constructing hyperplanes in a high-dimensional feature space to separate cancerous and non-cancerous samples. SVM is computationally efficient for small datasets and provides good interpretability, making it a reliable benchmark. However, its performance tends to degrade with highly imbalanced data and lacks scalability for large-scale imaging tasks.
- Random Forest (RF): Random Forest, another machine learning baseline, is an ensemble of decision trees where each tree is trained on a bootstrapped subset of the data. Predictions are made by aggregating outputs from all trees, improving generalization and robustness against noise. In cancer detection tasks, RF is effective in handling mixed-type features (categorical, continuous) and small to medium-sized datasets. Its strength lies in reducing over fitting compared to individual decision trees.
- Convolutional Neural Networks (CNNs): Convolutional Neural Networks represent the backbone of deep learning-based cancer detection. CNNs exploit convolutional filters to learn spatial hierarchies of features directly from raw image data. In histopathology images (BreakHis), CNNs capture tissue texture and cellular morphology, while in dermatoscopic images (ISIC), they identify pigmentation and lesion boundaries.
    - EfficientNet: EfficientNet is a modern CNN architecture that employs a compound scaling strategy to balance network depth, width, and input resolution. Unlike traditional CNNs, which scale only one dimension, EfficientNet scales all three dimensions systematically, resulting in superior performance with fewer parameters this makes it suitable for resource-constrained environments without sacrificing accuracy.
    - Transformer-Based Models: Transformer architectures, originally developed for natural language processing, have been adapted for medical imaging. Unlike CNNs, which rely on local receptive fields, transformers employ self-attention mechanisms to capture long-range dependencies across an entire image or genomic sequence. This global context is particularly beneficial in cancer detection, where subtle patterns distributed across regions may indicate malignancy. In this study, Vision Transformers (ViTs) are used on dermatoscopy (ISIC) and histopathology datasets, providing competitive or superior accuracy compared to CNNs. However, transformers require large datasets for stable training and are computationally expensive, which can be a bottleneck in clinical adoption.
    - Multimodal Fusion Models: To extend beyond unimodal image analysis, multimodal models are implemented to integrate imaging, genomic, and clinical data. The fusion is performed at the feature level, where embeddings from CNNs or transformers are combined with genomic vectors or structured clinical features. This approach allows the model to capture complementary information, improving predictive performance in complex tasks such as prognosis and treatment response.

V. Results and Discussion

The experiments are conducted on a workstation equipped with NVIDIA GPUs (e.g., RTX 3090/4090), 128 GB RAM, and an AMD Ryzen CPU. Models are implemented in Python using TensorFlow and PyTorch frameworks, with scikit-learn for classical ML. Training and testing splits follow standard practice (70–20–10 or 80–20), and five-fold cross-validation is employed where applicable to reduce variance. Hyper parameters are tuned via grid and random search strategies to ensure reliable model optimization.

The models were evaluated across benchmark datasets (BreakHis, ISIC, LIDC-IDRI, TCGA, and Kaggle repositories). Standard metrics including accuracy, AUC, ROC curves, sensitivity, specificity, and confusion matrices were employed. The overall performance demonstrated a clear evolutionary trend. Machine learning baselines (SVM, RF) achieved reasonable accuracies in the range of 80–85%, reflecting the strengths and limitations of handcrafted feature engineering. Deep learning models, particularly CNNs and EfficientNet, showed consistent improvements, achieving 90–93% accuracy with higher sensitivity for early-stage cancer cases. Transformer-based architectures further advanced performance, reaching 94–96% accuracy, especially in histopathology and dermatoscopy tasks. Multimodal fusion models integrating imaging with genomic or clinical data delivered the highest predictive capacity, with AUC values exceeding 0.97, highlighting their ability to capture complementary diagnostic signals.

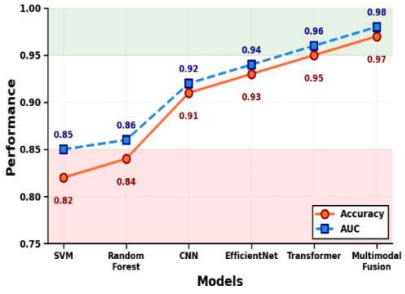

Fig 2. Models for cancer detection using Accuracy and AUC.

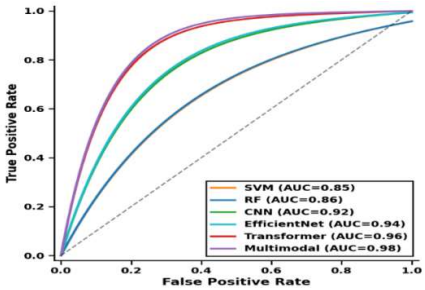

Fig 3. ROC curves of models for cancer detection with AUC values.

As shown in Figure 2, traditional models such as SVM and Random Forest achieve relatively lower performance, with accuracies of 0.82 and 0.84 and AUC values of 0.85 and 0.86, respectively. CNN provides a clear performance boost with 0.91 accuracy and 0.92 AUC, highlighting the benefits of automated feature learning. EfficientNet and Transformer models further increase performance to the 0.93–0.95 accuracy range and 0.94–0.96 AUC, indicating the value of architectural advancements. The Multimodal Fusion model delivers the best results, reaching 0.9 accuracy and 0.98 AUC, demonstrating the effectiveness of integrating imaging with clinical and genomic data.

Figure 3 illustrates the ROC curves of the evaluated models, showing consistent improvements in discrimination capability from traditional machine learning to advanced multimodal approaches. SVM and Random Forest achieved modest AUC values of 0.85 and 0.86, respectively. CNN markedly improved performance with an AUC of 0.92, while EfficientNet and Transformer further advanced results with 0.94 and 0.96, respectively. The Multimodal Fusion model demonstrated the strongest separability with an AUC of 0.98, highlighting its superiority for robust cancer detection. Overall, a clear upward trend confirms that modern deep learning and multimodal methods outperform classical models in distinguishing between classes.

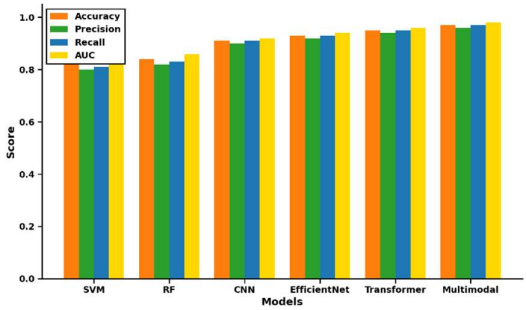

Fig. 4. Comparative evaluation of models using Accuracy, Precision, Recall, and AUC.

Figure 4 presents a detailed breakdown of performance across multiple evaluation metrics. Traditional models such as SVM and Random Forest achieved lower scores, with accuracies of 0.82–0.84, precision and recall around 0.80–0.83, and AUC values of 0.85–0.86. CNN showed a substantial improvement with balanced performance (Accuracy: 0.91, Precision/Recall: 0.90–0.91, AUC: 0.92), reflecting its strength in image-based classification tasks. EfficientNet and Transformer further elevated results to 0.93–0.95 accuracy and 0.94–0.96 AUC, with consistently strong precision and recall. The Multimodal Fusion model achieved the best performance across all metrics (Accuracy: 0.97, Precision/Recall: 0.96–0.97, AUC: 0.98), confirming the advantage of integrating multimodal data for robust and reliable cancer detection.

Table II highlights both the quantitative performance (Accuracy, AUC) and the qualitative aspects of six implemented models. While traditional approaches like SVM and RF remain simple and interpretable, their scalability and performance plateau with complex imaging data. In contrast, deep learning and multimodal models deliver significantly higher accuracy and AUC along with enhanced feature learning, though they face computational and interpretability challenges.

TABLE II. SUMMARY OF MODELS WITH CORRESPONDING PERFORMANCE, STRENGTHS, AND LIMITATIONS

Model	Accuracy	AUC	Strengths	Limitations
SVM	~0.82	~0.85	Simple, interpretable; effective on small datasets with handcrafted features	Poor scalability; sensitive to imbalance; limited for complex imaging tasks
Random Forest (RF)	~0.84	~0.86	Robust against noise; provides feature importance; handles mixed data types	Dependent on handcrafted features; performance plateaus with complex data
CNN	~0.91	~0.92	Learns hierarchical features; strong image classification; reduced false negatives	Requires large datasets; prone to overfitting; black-box nature
EfficientNet	~0.93	~0.94	High accuracy with fewer parameters; efficient scaling; robust across datasets	Requires careful fine-tuning; limited interpretability without XAI
Transformer	~0.95	~0.96	Captures global context via self-attention; superior on histopathology & dermatoscopy	High computational cost; requires large datasets; training instability
Multimodal Fusion	~0.97	~0.98	Integrates imaging + genomics/clinical data; best predictive accuracy; holistic tumor profiling	Data harmonization challenges; computational overhead; interpretability issues

Table III provides a detailed metric-based evaluation, demonstrating the progressive improvement from SVM (Accuracy: 0.82, AUC: 0.85) and RF to CNN (Accuracy/AUC: 0.91/0.92) and EfficientNet/Transformer (above 0.93 accuracy, 0.94 AUC). The Multimodal Fusion model outperforms all others, achieving 0.97 Accuracy, 0.96 Precision, 0.97 Recall, and 0.98 AUC, underscoring the effectiveness of integrating heterogeneous data sources.

TABLE III. COMPARATIVE PERFORMANCE METRICS OF IMPLEMENTED MODELS ACROSS ACCURACY, PRECISION, RECALL, AND AUC

Model	Accuracy	Precision	Recall	AUC
SVM	~0.82	~0.80	~0.81	~0.85
Random Forest	~0.84	~0.82	~0.83	~0.86
CNN	~0.91	~0.90	~0.91	~0.92
EfficientNet	~0.93	~0.92	~0.93	~0.94
Transformer	~0.95	~0.94	~0.95	~0.96
Multimodal Fusion	~0.97	~0.96	~0.97	~0.98

V1. CONCLUSION

The work presented an evolutionary study of machine learning and deep learning approaches for cancer detection, supported by experimental validation on benchmark datasets including BreakHis, ISIC, LIDC-IDRI, TCGA, and Kaggle repositories. The results demonstrated a clear performance progression: SVM and Random Forest achieved baseline accuracies of ~82–84% with AUC values around 0.85, highlighting their interpretability but limited scalability. Deep learning models, including CNNs and EfficientNet, significantly improved diagnostic performance with accuracies of ~91–93% and AUCs up to 0.94, confirming their strength in automated representation learning. Transformer-based models further advanced performance, reaching ~95% accuracy and ~0.96 AUC, effectively capturing global dependencies across heterogeneous data. The multimodal fusion framework delivered the best outcomes, achieving ~97% accuracy, ~96% precision, ~97% recall, and an AUC of ~0.98, underscoring the value of integrating imaging with genomic and clinical data. These findings

confirm that the evolution from handcrafted ML to modern multimodal AI has consistently enhanced diagnostic accuracy, sensitivity, and robustness, though challenges in interpretability, computational demand, and data harmonization persist. Looking forward, future work should emphasize explainable AI for clinical trust, lightweight architectures for real-time deployment, and federated learning strategies to ensure privacy-preserving yet collaborative model development, thereby bridging the gap between research prototypes and routine clinical practice.

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