# Artificial Intelligence-Driven Early Detection of Sarcoma Using Radiogenomics and Clinical Data Fusion

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**Abstract**

**Purpose:** Sarcoma, a rare and heterogeneous group of cancers, poses significant challenges for early detection due to its deep tissue origin, complex biology, and genetic diversity. The integration of artificial intelligence (AI), specifically machine learning (ML) and deep learning (DL), offers a promising pathway to improve diagnostic accuracy and enable personalized treatment strategies.

**Methods:** We present a multimodal AI-driven framework for early sarcoma detection using datasets from The Cancer Imaging Archive (TCIA) and The Cancer Genome Atlas (TCGA-SARC). Convolutional Neural Networks (CNNs) were applied for imaging feature extraction, while genomic data were processed with classical ML models. A multimodal fusion model integrated imaging, genomic, and clinical metadata with attention mechanisms and Grad-CAM explainability.

**Results:** The multimodal model achieved superior performance (Accuracy: 94.3%, AUC-ROC: 0.951) compared to single-modality approaches. Grad-CAM visualizations highlighted tumor regions, enhancing interpretability.

Conclusion: This study demonstrates the feasibility of integrating multimodal AI techniques for sarcoma detection, laying the foundation for precision diagnostics, prognosis prediction, and therapy optimization.

Keywords: Sarcoma, Soft Tissue Sarcoma, Artificial Intelligence, Machine Learning, Deep Learning, Detection

## Introduction

Sarcomas are rare malignant tumors arising from mesenchymal tissues, accounting for 1–2% of adult malignancies. Their heterogeneity, deep anatomical location, and over 70 histological subtypes complicate early detection. Current diagnostic methods often lead to late-stage diagnosis. AI-driven approaches, proven in breast and lung cancer detection, remain underexplored in sarcomas due to data scarcity and subtype variability [1]. This study introduces a multimodal AI framework integrating imaging, genomic, and clinical data for sarcoma detection.

**Related Work**

AI applications in oncology have advanced significantly, particularly in breast, lung, and prostate cancers. CNNs achieve dermatologist-level performance in skin lesion classification, while radiomics and radiogenomics enable tumor characterization and genotype-phenotype correlations. However, sarcoma research is limited due to rare datasets and subtype heterogeneity. Some studies explored MRI radiomics and genomic prediction for sarcomas, but most lack multimodal integration and explainability. Our study addresses these gaps with an interpretable AI-based multimodal system [2-3].

**Materials and Methods**

Dataset: 300 patient records combining TCIA imaging, TCGA-SARC genomics, and clinical metadata.

Preprocessing: Imaging resized to 224x224, normalized, and augmented. Genomics filtered, normalized with log2(FPKM+1), and reduced via PCA/RFE. Clinical data imputed and one-hot encoded.

Model Development: CNNs for imaging, ML classifiers (RF, SVM) for genomics, and fusion with attention mechanisms in a deep neural network. Training used Adam optimizer (lr=0.001), cross-entropy loss, and early stopping.

Evaluation: 10-fold stratified CV, metrics included Accuracy, Precision, Recall, F1, and AUC-ROC. Grad-CAM and SHAP were applied for interpretability[4-6].

**Results and Discussion**

CNN (Imaging): 91.2% Accuracy, AUC 0.927.
Random Forest (Genomics): 89.0% Accuracy, AUC 0.901.
SVM (Genomics): 87.5% Accuracy, AUC 0.887.
Multimodal Fusion: 94.3% Accuracy, AUC 0.951.

**Discussion**

The findings of this study demonstrate the feasibility and potential of an AI-driven multimodal approach for the early detection of sarcoma, addressing a critical gap in current diagnostic methodologies. By leveraging imaging data, genomic profiles, and clinical metadata, the proposed framework achieved a superior performance compared to single-modality models, showcasing the strength of integrative data analysis in rare cancer detection.

**Significance of Multimodal Integration**

Single-modality limitations: The CNN-based imaging model, though effective (91.2% accuracy), was limited in distinguishing between subtypes with subtle morphological differences. Similarly,

genomics-only models (e.g., Random Forest with 89.0% accuracy) struggled with the inherent noise and high dimensionality of molecular data.
Synergistic advantage: By fusing features from imaging, genomics, and clinical data, the multimodal model captured a more holistic representation of the tumor phenotype and genotype, leading to a significant increase in accuracy (94.3%) and AUC-ROC (0.951).
This outcome validates existing literature emphasizing the value of radio genomics in oncology, but our work uniquely applies this integration specifically to sarcoma, a largely underserved domain in AI research.

**Attention Mechanisms & Model Explainability**

The use of an attention mechanism in the fusion layer allowed the model to prioritize modality-specific features, dynamically adjusting the importance of imaging vs. genomic vs. clinical variables per patient. Grad-CAM visualizations further enhanced model transparency, offering interpretability at the image level by highlighting tumor-relevant regions. This is crucial for clinical acceptance, as explainable AI can bridge the gap between black-box models and physician trust.

**Challenges and Limitations**

While the study presents promising results, several challenges remain:
Data scarcity: Sarcoma datasets remain limited in size and diversity. The 300 patient samples used, though substantial for sarcoma research, are still small by deep learning standards.
Subtype imbalance: Certain rare sarcoma subtypes were underrepresented, potentially affecting model generalization.
Data heterogeneity: Variability in imaging protocols, sequencing platforms, and clinical data recording could introduce biases.
External validation: The model's performance needs to be tested on independent, multi-institutional cohorts to assess its real-world applicability.

**Clinical Implications**

An AI-based early detection tool for sarcoma can significantly reduce diagnostic delays, allowing for earlier intervention and improved patient outcomes. The framework has potential applications in clinical decision support systems (CDSS), assisting radiologists and oncologists in differential diagnosis. Additionally, the model can be extended to assist in treatment response prediction and prognostic assessments, paving the way for precision oncology in sarcoma care.

**Conclusion and Future Work**

This study presents a novel, AI-driven multimodal pipeline for the early detection of sarcoma, integrating radiological imaging, genomic data, and clinical information. The model demonstrated:
Superior predictive performance (94.3% accuracy, 0.951 AUC-ROC)

Enhanced interpretability through Grad-CAM visualizations
Improved feature prioritization via attention mechanisms
These findings underscore the potential of AI in addressing diagnostic challenges in rare cancers like sarcoma, which are often neglected in mainstream AI oncology research.

**Future Work**

Building upon this foundation, future directions will focus on:
Dataset Expansion: Collaborating with global sarcoma registries and consortia to enhance data diversity and subtype representation.
Real-time Clinical Deployment: Developing user-friendly interfaces and integrating the model into hospital information systems for real-world application.
3D Imaging and Temporal Analysis: Incorporating 3D volumetric imaging data and longitudinal follow-ups to improve predictive accuracy.
Explainable AI Enhancements: Exploring more advanced interpretability methods (e.g., SHAP, LIME) to further elucidate model decisions.
Extension to Prognosis and Therapy Prediction: Expanding the model's application to predict patient prognosis, metastasis risk, and treatment responses.

By addressing these future directions, this work aims to contribute significantly to the field of AI-assisted precision medicine in rare cancers, ultimately improving sarcoma patient outcomes.

**References**

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