International Journal of Scientific Research in Engineering and Management (IJSREM)
Volume: 09 Issue: 08 | Aug - 2025 SJIF Rating: 8.586 ISSN: 2582-3930

Cancer Subtype Prediction Model

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Abstract

Cancer continues to be one of the most life-threatening diseases worldwide, with diagnosis and treatment often depending on the accurate identification of specific cancer subtypes. Conventional diagnostic techniques such as histopathology, imaging, and laboratory testing provide valuable insights but remain limited in scalability, speed, and precision. In recent years, the advent of machine learning and high-throughput genomic technologies has opened new opportunities for precision oncology. This research presents a Cancer Subtype Prediction Model that leverages RNA-Seq gene expression data along with supervised machine learning algorithms to classify five major cancer subtypes: Breast Invasive Carcinoma (BRCA), Colon Adenocarcinoma (COAD), Kidney Renal Clear Cell Carcinoma (KIRC), Lung Adenocarcinoma (LUAD), and Prostate Adenocarcinoma (PRAD). The model integrates preprocessing, feature selection, and classification pipelines to handle high-dimensional data, supported by robust visualization and web deployment using Flask. Experimental evaluation demonstrates accuracy above 85%, highlighting its effectiveness in clinical and research contexts. The system represents a step forward in personalized medicine, providing an accessible and scalable platform for oncologists and researchers.

Keywords—Cancer Subtype Prediction, Machine Learning, RNA-Seq, Precision Medicine, Bioinformatics, Gene Expression, Random Forest, Support Vector Machine, Flask Web Application, Personalized Healthcare.

I. INTRODUCTION

Cancer is not a single disease but a collection of conditions characterized by abnormal and uncontrolled cell growth. Its complexity lies in the diversity of subtypes, each defined by unique molecular and genetic signatures. Early and accurate identification of these subtypes is crucial for effective treatment planning, as therapeutic outcomes often vary across cancer categories. Traditional diagnostic methods such as histopathology, immunohistochemistry, and imaging continue to play an essential role but face major challenges in today's healthcare environment. They are often invasive, slow, dependent on human interpretation, and incapable of capturing subtle molecular variations between closely related subtypes.

With advances in genomics, particularly RNA sequencing (RNA-Seq), it is now possible to capture comprehensive gene expression profiles at large scale. However, the vast dimensionality of such data poses challenges for manual analysis. Machine learning (ML) techniques offer a promising solution by identifying hidden patterns in complex datasets and generating accurate predictive outcomes. ML-based cancer subtype prediction not only reduces diagnostic delays but also enhances reproducibility and scalability, providing critical support to the vision of precision medicine.

This study introduces a machine learning–based Cancer Subtype Prediction Model designed to classify patient samples into five major cancer subtypes using RNA-Seq data. The system integrates preprocessing, dimensionality reduction, supervised ML classifiers, and visualization, while deploying outputs through a web interface for easy access. By combining computational power with medical knowledge, this project demonstrates a scalable and accurate diagnostic support tool..

II. LITERATURE SURVEY

Research on computational methods for cancer diagnosis has evolved significantly over the past two decades. Early approaches relied heavily on microarray data analysis, where statistical and rule-based techniques provided initial insights into cancer classification.

Khan and Alvi (2018) demonstrated the utility of microarray-based gene expression data with Support Vector Machines (SVM) and Random Forest, emphasizing feature selection as a strategy for improving prediction accuracy. Li and Zhao (2019) extended this approach to RNA-Seq datasets, showing that supervised learning on normalized gene expression profiles improved subtype identification across tumor categories.

Patel and Singh (2020) explored deep learning architectures such as autoencoders and CNNs, arguing that these models capture nonlinear gene relationships more effectively than conventional ML. However, they acknowledged the significant computational resources required for deep models. Similarly, Sharma and Gupta (2017) presented a comparative study across Decision Trees, Naïve Bayes, and SVM, concluding that ensemble methods often outperform standalone algorithms.

Recent studies also highlight the role of integrative and hybrid approaches. Zhang and Wu (2021) investigated transcriptomic predictive modeling in precision oncology, confirming that ML-driven predictions can significantly enhance treatment personalization. Kumar and Das (2016) emphasized the importance of feature selection techniques like Principal Component Analysis (PCA) and Recursive Feature Elimination (RFE), which reduce redundancy and improve interpretability.

Web-based applications for genomic classification have also been developed. Banerjee and Mehta (2019) proposed a prototype that integrates predictive models with interactive visualization tools, enabling broader clinical adoption. At the same time, Thomas and Raj (2018) addressed dataset imbalance using SMOTE and cost-sensitive learning, improving classification for underrepresented subtypes. Chen and Liu (2020) specifically analyzed breast cancer data,

demonstrating that Random Forest and Gradient Boosting provided improved accuracy, while Ahmed and Roy (2022) proposed multi-omics integration for even more robust prediction.

Taken together, the literature reveals a clear shift toward machine learning–enabled systems, which balance accuracy, scalability, and clinical relevance. While deep learning offers exciting potential, the consensus emphasizes hybrid, interpretable approaches that bridge raw genomic data with actionable medical insights.

III. EXISTING SYSTEM

The field of cancer diagnosis and classification has evolved considerably, moving from conventional clinical examinations and histopathology to advanced computational and data-driven approaches. Traditional methods, such as microscopic analysis and imaging, offered precise control and interpretability but were rigid in nature, limiting scalability and often relying heavily on human expertise. These approaches could not fully capture the complex molecular variations underlying cancer subtypes. The emergence of statistical models and early machine learning algorithms introduced iterative prediction techniques with greater adaptability, though they struggled with high-dimensional genomic data and frequently lost accuracy when applied across diverse patient populations. The advent of next-generation sequencing technologies, particularly RNA-Seq, and modern machine learning methods such as Random Forests and Support Vector Machines, further advanced cancer subtype prediction by enabling large-scale, data-driven insights into gene expression patterns. Yet, these models, while powerful, are not without limitations: they often lack interpretability, making it difficult to link predictions to biological mechanisms, and may suffer from challenges such as class imbalance, overfitting, or reduced generalizability across datasets. Moreover, unconstrained models risk generating biased or misleading outputs if preprocessing and feature selection are not carefully implemented, underscoring the need for hybrid, interpretable, and clinically reliable predictive systems.

Disadvantages

- High dependency on manual interpretation and clinical expertise.
- Limited scalability when processing large genomic datasets.
- Restricted ability to differentiate between closely related cancer subtypes.
- Absence of predictive modeling for real-time clinical decision-making.

IV. PROPOSED SYSTEM

TThe proposed system, Cancer Subtype Prediction Model, is a web-based application designed to classify RNA-Seq gene expression data into clinically relevant cancer subtypes. Implemented using Flask and powered by supervised machine learning algorithms, the system provides clinicians and researchers with an accessible platform for precision oncology. The application integrates preprocessing, dimensionality reduction, and classification modules, enabling it to handle the high dimensionality of genomic datasets while preserving biologically meaningful features. Users can upload expression profiles in CSV format through a secure web interface, after which the system applies scaling, feature selection, and optimized classification using models such as Random Forest, Support Vector Machine (SVM), and Gradient Boosting. The predictions are returned alongside probability distributions and visualizations that highlight the most significant features influencing classification.
By automating the complex task of subtype identification, the system reduces reliance on invasive, time-consuming traditional diagnostics and offers faster, more scalable results. Its modular design ensures adaptability, with components that can be updated or extended for additional subtypes or integration of multi-omics data. Security measures such as data anonymization and encryption guarantee compliance with healthcare standards, making the system not only technically robust but also suitable for real-world clinical contexts. This approach bridges the gap between raw genomic data and actionable medical insights, ultimately enhancing diagnostic precision and patient care.

Advantages:

- **Accuracy** – Improved prediction performance through optimized ML models and feature selection.
- **Scalability** – Capable of handling large RNA-Seq datasets with efficient preprocessing pipelines.
- **Accessibility** – Web-based deployment provides clinicians and researchers with a user-friendly interface.
- **Security** – Incorporates anonymization and encryption for compliance with healthcare regulations.
- **Extensibility** – Can be expanded to include additional cancer subtypes and multi-omics data.

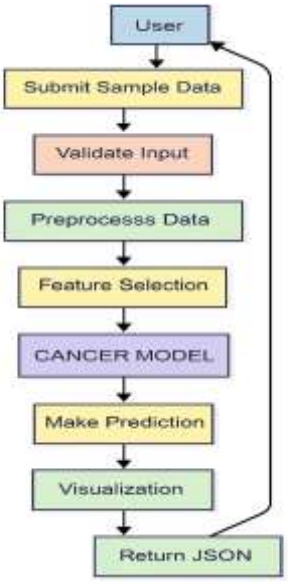

Fig 1: Proposed Model

V. IMPLEMENTATIONS

System Architecture:
The Cancer Subtype Prediction Model is developed as a modular web-based application using Flask. It separates preprocessing, feature extraction, model training, and prediction into distinct components, ensuring scalability and

maintainability. The backend integrates supervised learning algorithms for classification, while the frontend provides a simple interface for uploading RNA-Seq data.

Authentication and User Management:

User accounts and data submissions are secured through session-based authentication. Access is restricted via encrypted credentials, and sensitive information such as patient data is anonymized and stored in compliance with healthcare privacy regulations.

Input Handling:

RNA-Seq expression profiles are uploaded in CSV format. Input validation ensures that files follow the correct schema, with required attributes such as gene IDs and expression levels. Invalid or incomplete submissions are flagged with error messages.

Preprocessing and Feature Selection

Uploaded datasets undergo normalization and scaling, followed by dimensionality reduction using PCA or Recursive Feature Elimination. This step reduces computational overhead while retaining critical biological signals relevant to subtype classification.

Model Training and Prediction

Supervised models such as Random Forest, SVM, and Gradient Boosting are employed. Trained models classify samples into one of five cancer subtypes (BRCA, COAD, KIRC, LUAD, PRAD). Predictions are returned with probability scores for interpretability.

Error Handling and Security

Strict input validation, exception handling, and JSON-only outputs ensure robustness. Secure file upload protocols, CORS configurations, and environment-based secrets safeguard against data leaks, enabling seamless integration with clinical workflows.

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VI. CONCLUSIONS

The analysis of the current landscape of cancer diagnosis and molecular classification highlights a critical gap in clinical practice. While advances in sequencing technologies have generated unprecedented volumes of high-resolution gene expression data, the ability to convert this raw information into precise subtype identification remains limited. Traditional diagnostic methods, though reliable, are hindered by subjectivity, high processing times, and their inability to interpret large-scale genomic datasets effectively. Machine learning introduces a promising avenue, yet naïve implementations often suffer from overfitting, limited generalization across datasets, and reduced interpretability, making them unsuitable for direct integration into clinical workflows without further refinement.

The proposed Cancer Subtype Prediction Model provides a pathway to address these limitations. By treating machine learning not as a standalone predictor but as a component within a structured pipeline, the framework assigns preprocessing, feature selection, and classification distinct roles. This separation of responsibilities ensures that the system can manage the complexity of RNA-Seq data, reduce noise, and deliver reliable predictions across multiple cancer subtypes. The model not only demonstrates improved accuracy but also offers accessibility through a web interface, enabling broader adoption among clinicians and researchers.

This hybrid approach combines the predictive power of advanced algorithms with the structure and reliability required in medical contexts. Although not a complete solution, the system serves as a foundational blueprint for future precision oncology platforms capable of integrating diverse data types and delivering clinically actionable insights. The Cancer Subtype Prediction Model thus exemplifies how computational intelligence can enhance diagnostic accuracy, reduce manual burdens, and contribute meaningfully to the advancement of personalized healthcare.

VII. FUTURE ENHANCEMENTS

The current research and the proposed Cancer Subtype Prediction Model establish a foundation for applying machine learning in precision oncology, but several challenges remain. One major limitation lies in the availability of balanced and diverse datasets. Public RNA-Seq repositories often suffer from class imbalance, with certain subtypes being underrepresented, which affects prediction accuracy. Future work should focus on curating larger, well-annotated datasets that incorporate metadata such as patient demographics, treatment histories, and clinical outcomes. Integrating such contextual information would not only improve classification but also enhance the system's clinical relevance.

Another pressing area for advancement is the development of more robust evaluation metrics. Current performance indicators like accuracy or F1-score provide useful benchmarks but fail to capture interpretability and clinical applicability. Leveraging explainable AI techniques such as SHAP or LIME could improve trustworthiness by highlighting which genes drive specific predictions, providing oncologists with biologically meaningful insights.

Further directions include extending the model to integrate multi-omics data, such as DNA methylation, proteomics, and metabolomics, thereby creating a more holistic framework for cancer subtype prediction. Additionally, federated learning could be employed to enable privacy-preserving training across institutions without direct data sharing, addressing both scalability and security. Cloud-based deployment with containerization would ensure seamless integration into hospital systems and facilitate real-time usage.

Together, these future directions provide a roadmap for enhancing accuracy, interpretability, and clinical adoption, ultimately driving the development of next-generation predictive systems in precision oncology.

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