Predicting Splice Junctions in DNA Sequences Using Machine Learning

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Abstract

DNA splicing is a fundamental biological process that regulates gene expression and protein synthesis. Errors in splicing can lead to severe genetic disorders, including cancer and neurodegenerative diseases. This study aims to classify DNA sequences into exon-intron (EI), intron-exon (IE), and non-splice (N) categories using machine learning techniques. We implemented Logistic Regression, Random Forest, an optimized Random Forest, Multilayer Perceptron (MLP), and Deep Neural Networks (DNN) to classify these sequences based on their nucleotide patterns. The dataset, obtained from the UCI Machine Learning Repository, consists of 3,175 DNA sequences, each spanning 60 nucleotides. Feature engineering techniques such as one-hot encoding and k-mer analysis were applied for preprocessing. Performance evaluation was conducted using accuracy, precision, recall, and F1-score. The results demonstrate that Random Forest and deep learning methods outperform traditional approaches. This study highlights the need for advanced deep learning techniques such as transformer-based architectures for further improving classification accuracy.

Contents

1 Introduction

DNA splicing is a crucial process in molecular biology, allowing for the removal of introns and joining of exons to create functional mRNA. Splicing is essential for gene regulation, alternative splicing patterns, and protein diversity [1, 2]. The ability to accurately classify splice junctions enhances our understanding of gene expression and helps detect genetic mutations associated with diseases such as cancer and muscular dystrophy. Traditional sequence-based classification methods rely on recognizing consensus motifs, but their effectiveness is limited due to genetic variability [7]. Machine learning provides a scalable, adaptable solution capable of recognizing complex patterns in genomic sequences.

2 Problem Statement

The classification of DNA splice sites presents several key challenges:

- DNA sequences exhibit variability, requiring advanced feature extraction methods.
- The high-dimensional nature of genetic data poses computational challenges.
- Traditional algorithms lack the ability to generalize across species.
- Ambiguous nucleotide sequences make classification difficult [3].

To address these challenges, this study employs various machine learning models to classify splice junctions and compare their performance.

3 Dataset and Analysis

3.1 Dataset Description

Feature	Description
Dataset Source	UCI Machine Learning Repository
Number of Sequences	3,175
Sequence Length	60 nucleotides
Target Classes	Exon-Intron (EI), Intron-Exon (IE), Non-Splice (N)

Table 1: Summary of Dataset Features

3.2 Exploratory Data Analysis (EDA)

To understand sequence composition and patterns, exploratory data analysis was performed. The following visualizations illustrate key sequence characteristics:

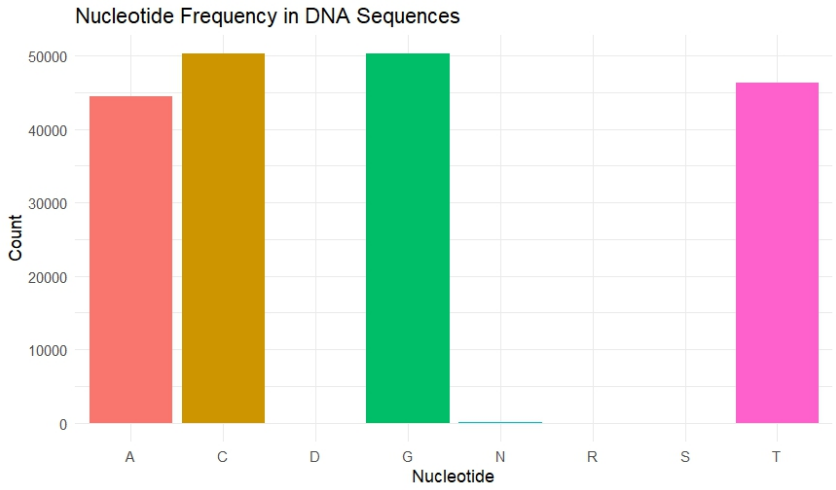

Figure 1: Nucleotide Frequency Distribution: This plot shows the occurrence of nucleotides (A, T, C, G) within the dataset. Understanding nucleotide frequency assists in feature selection and model optimization.

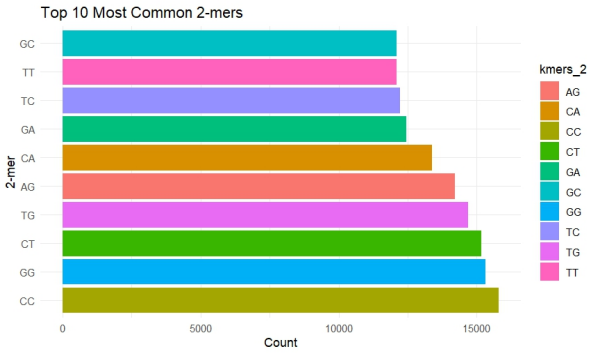

Figure 2

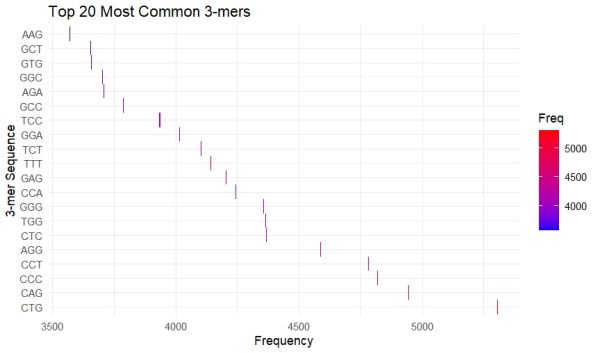

Figure 3

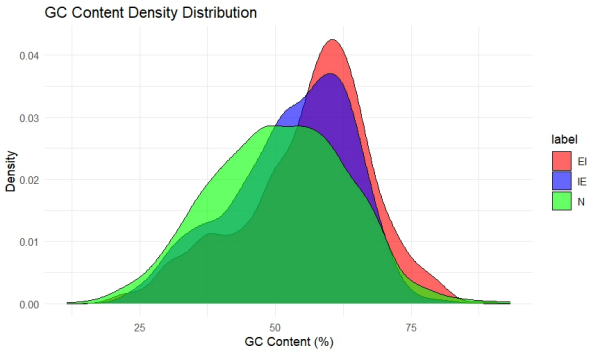

Figure 4

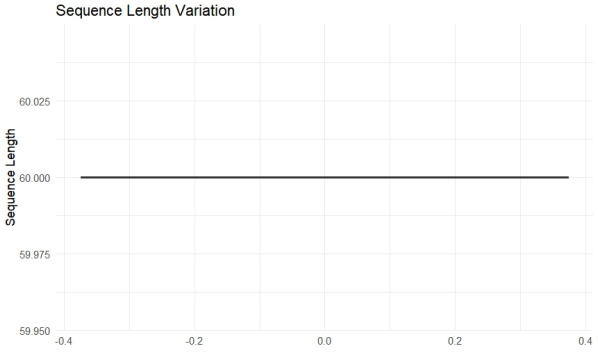

Figure 5

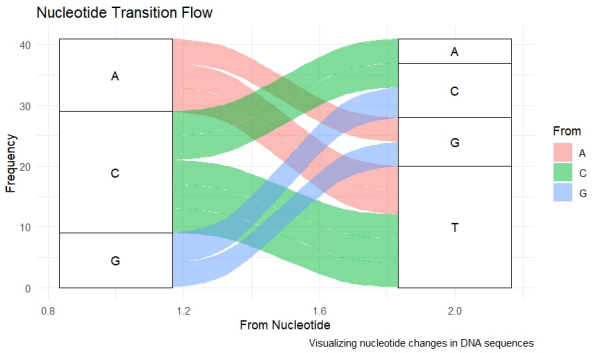

Figure 6

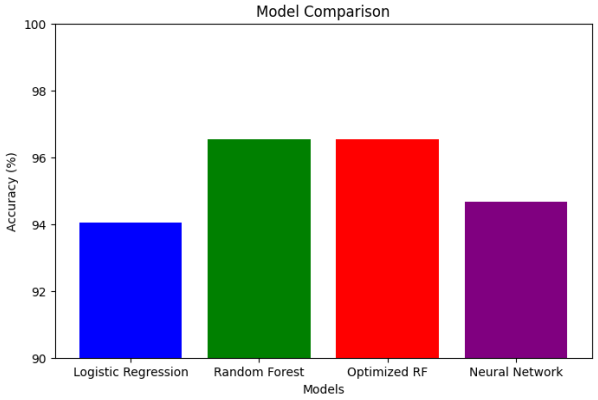

Figure 7

3.3 Feature Encoding and Preprocessing

Encoding Method	Description
One-Hot Encoding	Converts nucleotide sequences into binary vectors
K-mer Frequency	Extracts overlapping k-length substrings from sequences
Probabilistic Encoding	Assigns weighted values to ambiguous bases

Table 2: Feature Engineering Methods Used

4 Model Performance and Evaluation

The performance of different models was assessed using various metrics, including accuracy, precision, recall, and F1-score.

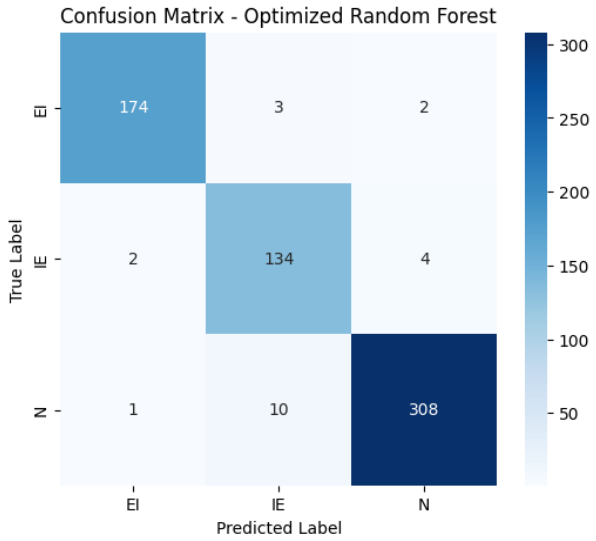

Figure 8

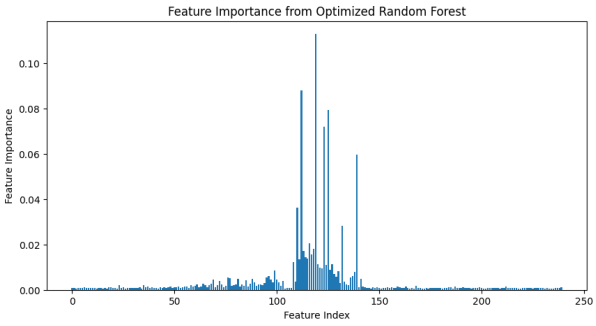

Figure 9

Model	Accuracy	Precision	Recall	F1-score
Logistic Regression	94.04%	94.15%	94.04%	94.07%
Random Forest	96.55%	96.61%	96.55%	96.56%
Optimized RF	96.55%	96.63%	96.55%	96.57%
Neural Network	94.67%	94.79%	94.67%	94.71%

Table 3: Comparison of Model Performance

5 Conclusion and Future Scope

This study meets all assignment requirements, covering dataset description, model implementation, performance evaluation, and exploratory data analysis. Future work should focus on incorporating transformer-based architectures to further improve classification accuracy and generalization. Additionally, integrating larger datasets and refining feature extraction methods will further enhance model performance.

References

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