A Comprehensive Review of Novel AI Techniques and Applications in Bioinformatics

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Abstract- Integrating Artificial Intelligence (AI) models into bioinformatics opens new avenues in biological data analysis and interpretation. The current study follows PRISMA guidelines for the search strategy, and the databases covered include PubMed, Embase, and Google Scholar for keywords that focus on studies published between 2017 and 2024. As for the aimed bioinformatics domains, we explored the uses of AI methodologies such as Machine Learning (ML), Deep Learning (DL), and Natural Language Processing (NLP) in the broad bioinformatics field. Other applications of this paradigm are genome sequence analysis, 2D/3D protein structure folding and prediction, systems biology, customized medicine for individuals, drug discovery, medical image analysis, signals and pathways processing, clinical data analysis, and biomedical text mining. AI systems have effectively addressed complex biological problems, from drug development to personalized medicine, protein structure prediction, and protein folding. In summary, this paper examines the rapidly changing field of AI tools and algorithms and their integration with bioinformatics. It highlights their critical function in accelerating biomedical research, simplifying data interpretation, and stimulating advancements.

Keywords- Machine Learning, Deep Learning, Natural Language Processing, Bioinformatics, Biomedical

I. INTRODUCTION

Bioinformatics plays a critical role in bridging the gap between various aspects of computational and biological data analysis tasks. It reveals latent patterns and significant insights within large complex datasets and offers the necessary framework and techniques for analyzing, interpreting, and deriving valuable knowledge. A substantial shift in the development of bioinformatics marked the last decade. The methods of Artificial Intelligence (AI) have become a driving force in innovating several domains. With the increase in next-generation sequencing and other sophisticated technologies, biological data, or big data, is becoming a big challenge mainly because of the high complexity and time taken to analyze the data [1-3].

From past few years, the rapid growth of biological dataset generated through omics technologies, next generation sequencing, and biomedical images has surpassed the analytical capabilities of traditional bioinformatics tools. The rapid growth of data has created the demand for more intelligent, saleable, reliable, and robust system to uncover biological meaningful insights. The integration of AI technologies such as ML, DL, and NLP have emerged as potential solution to address these challenges enabling automated feature extraction, accurate prediction and pattern recognition [4-5].

Numerous machine learning algorithms such as Support Vector Machines (SVM), random forest, and neural networks are being ensembled when dealing with such problems in bioinformatics [6-9]. These advanced techniques enable researchers to handle complex data to predict meaningful patterns. Transcription Factor Binding Sites (TFBSs), protein structure analysis, and biomarker prediction are some of the key areas for implementing AI techniques [10-12]. A few impactful innovations in bioinformatics using AI techniques are disease diagnosis and personalized medicine [13]. The success of AlphaFold has highlighted the capability of deep leaning models with predict protein structure with high level precision, revolutionize the field of structural bioinformatics [14]. By leveraging the AI model such as DeepCRISPR and CRISPR-Net, researchers has improved the CRISPR-Cas9 applications by optimizing RNA design and off target analysis significantly improving the accuracy of genome editing [15]. In genomics AI is enabling the process of identifying disease related genes, regulatory motif annotation and predicting variant effect across diverse population [16]. However, current AI systems in bioinformatics still tempered by challenges such as data imbalance, model interpretability, limited in generalization, and ethical concerns including model biasness and data privacy [17]. This review provides an overview of the cutting-edge AI approaches in bioinformatics,

highlighting key advancements and limitations. We provide a detailed comparison of recent research, emerging trends, and future pathways, and identify significant research gaps that still need to be addressed in the current literature. In this current review, key categories and emerging technologies of bioinformatics applications have been summarized. AI tools are making an impactful contribution to enhancing research and data interpretation for forecasting innovation in biomedical sciences. Furthermore, Table 1 presents the impact of the AI application in bioinformatics.

Table 1: Impact of AI Applications in Bioinformatics

Domain	AI Application	Techniques/Tools	Impact
Genomics	Disease Classification	DeepVariant [18]	Improved accuracy over traditional methods
Proteomics	Protein structure prediction	AlphaFold [14]	Revolutionize 3D protein modeling
CRISPR/ Cas9	Off-target analysis	DeepCRISPR [19]	Improve precision in genome editing
Drug Discovery	Molecular docking	Graph Neural Networks (GNNs) [20]	Accelerated potential drug discovery
Personalized Medicine	Patient stratification	SVM, Random Forests [21]	Personalized health care solutions

In this research, PubMed, Embase, and Google Scholar were used for the search strategy to identify the relevant articles. Studies published from 2018 to 2024 were used. The exclusion of the studies that were identified when performing the search is well illustrated by the PRISMA flowchart in Fig. 1. Firstly, using the research strategy, 362 articles were identified. subsequently, after filtering the studies based on the titles, keywords, and abstracts, we disregarded irrelevant studies. Further screening was done by reviewing the full texts of the remaining studies and discarding conference and unpublished papers. Finally, fewer than 60 papers were found to be relevant and focused on AI and bioinformatics. The keywords used for search are "Artificial Intelligence", "Machine Learning", "Deep Learning", and others related to bioinformatics are "Genomic Analysis", "Protein Structure Prediction", "Drug Discovery", "Personalized Medicine", and "Biomedical Signal Processing". Finally, a manual search was conducted to retrieve the most relevant and potential research.

The rest of the paper is organized as follows: Section 2 discusses the related work relevant to the study. Section 3 explains the literature review of the research. Section 4 presented the evaluation parameter and section 5 demonstrated the discussion. Section 6 discusses the gap analysis and section 7 concludes the conclusion.

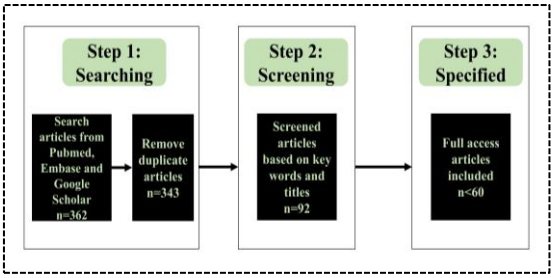

Fig. 1. PRISMA Flowchart

II. Related Work

The first-generation bioinformatics was essentially dependent on rule-based recognition, hands-on analysis, and statistical patterns for the biological information. Nevertheless, the increasing scale and complexity of biological sequential data generated from next-generation sequencing and high-throughput technology have made classical approaches insufficient to deal with the data generation speed and reliability [5]. The evolution from classical bioinformatics to smart is a progressive move in the bioinformatics field, mainly due to the enhanced employment of Artificial Intelligence (AI), Machine Learning (ML), and analysis of big data. The involvement of AI and ML in the study revealed that it improves the speed and precision of analyses, besides presenting unexplored possibilities for modeling intricate biological structures [22-23].

Complex sequential data processing, manual input of features, and rigidity are notable drawbacks of classical bioinformatics methods. Also, these classical approaches are limited when applied to noisy data, containing random fluctuations due to limited conditions of experiments, imprecision of measurement instruments, or biological variations. Noise can reduce the accuracy of the analyses made using classical methods [23]. There are numerous features of advanced bioinformatics that address these limitations: (1) Automated feature extraction and learning: Manual feature engineering is evaded automatically using AI techniques. Relationships and hidden patterns in the data can be forecast using this technique. (2) Easy adaptability: AI methods are highly adaptable and applicable to a wide range of biological data analysis tasks, including classification, clustering, and regression. (3) State-of-the-art performance: AI-driven approaches deliver top-tier performance with high accuracy and precision in various bioinformatics applications, such as analysis of protein structure, drug discovery, and gene expression [24-26].

All the constraints and problems that bioinformatics and AI encounter are solved effectively when the methodologies of the two are integrated. Due to the integration, a comprehensive inspection that utilization of the strengths of the two disciplines is achievable. Firstly, researchers could avoid

limitations in traditional computational methods by integrating new technologies, such as the integration of ML and DL models with bioinformatics. AI algorithms can therefore perform better in large-scale biological information, which extends the possibility of fine-tuning biological process modeling [27]. Secondly, the model's specificity, accuracy, and sensitivity can be enhanced using this combination. AI algorithms are effective in finding hidden complex patterns and associations with biological information that may be overlooked by conventional approaches. The integration of AI with bioinformatics helps researchers to build better models that are not only more accurate but also more stable and less likely to fail [28]. Bioinformatics can thus be used to find new patterns and relationships in biological networks. This concept assists in the recognition of biomarkers, drug targets, and disease pathways toward bringing efficient personalized medicine, as well as the development of drugs [29-30]. Table 2 presents a comprehensive overview of the studies, including datasets, AI models, and key findings to enhance the analytical value.

Table 2: AI Tool for the Bioinformatics Domain

Ref.	Bioinformatics domain	Data	AI Model used	Key findings
[31]	Omics	Genomic, Transcriptomic, and Microbiome Data	Random Forest, XGBoost, AdaBoost, KNN, LightGBM, Neural Networks, and SHAP	Achieved 94% AUC in multi-omics disease prediction
[32]	Omics	107 tumoral pancreatic samples from TCGA, 117,486 germline SNPs	Random Forest and Penalized Multinomial Logistic Regression	Achieved 91.4% classification accuracy
[33]	Omics	Proteomics dataset of Saccharomyces cerevisiae	Random Forest	Model facilitating functional annotation using classical machine learning.
[34]	Transcriptomics	DLPFC dataset	Hexagonal Convolutional Neural Network	Successfully segmented data using spatially structured inputs
[35]	Transcriptomics	mantle cell lymphoma, diffuse large B-cell lymphoma	Logistic regression, Bayesian network, CHAID, SVM, and Multilayer perceptron	Predicted gene expression-based disease by integrating multiple machine learning models.
[36]	Proteomics	Human and yeast dataset	Hybrid CNN + BiGRU - Attention-based model	Achieved 3.8% improvement in Max-F1 score over the state-of-the-art model SDN2GO
[37]	Proteomics	HMR195 dataset BG570 dataset	IHHO-CNN-LSTM and Improved Harris Hawk Optimization	Achieved 94.6% accuracy in predicting protein-coding regions.
[38]	Proteomics	271,160 sequences across 543 protein families	1D-CNN combined with BiLSTM and attention mechanisms	Achieved 98.3% F1-score outperforming traditional models.
[39]	Metagenomics	8,794 virulent, 4,992 antibiotic resistance, 18,296 non-pathogenic sequences	Support Vector Machine	Achieved accuracy of 81.72%.
[40]	Metagenomics	232 samples, 114 patients, and 118 controls.	Deep neural networks for phenotypic prediction	Predicted phenotype from metagenomic profiles
[41]	System Biology	Gene regulation datasets for ribosome-binding site sequences	DeepSwarm, AutoKeras, TPOT for architecture search and optimization	Proposed machine learning for sequence optimization.
[42]	Signal Processing	928 subjects aged 22-60 from New Delhi, Cape Town, Riga	Long short-term memory (LSTM), CNN, and Gramian angular field-CNN (GAF-CNN)	Utilized deep learning models for bio signal classification with more precision.
[43]	Biomedical Text Analysis	BioCreative II GM corpus	LSTM framework with dual-channel and sentence-level reading control gates	Proposed NLP-based deep learning framework for text and entity analysis.

III. LITERATURE REVIEW

Bioinformatics is a field that combines computational and biological data analysis techniques. Over the past few years, processing biological data such as omics, proteomics, and genomics has been challenging for researchers. After evaluating AI in bioinformatics, researchers can extract valuable insights from the datasets and provide promising solutions [44]. In this research, the numerous domains of bioinformatics and their integration with AI have been discussed and organized in separate sections. Fig. 2 demonstrates the integration of an AI model into bioinformatics. It shows how AI models such as ML, DL, and NLP map to the specific domains of bioinformatics, such as genomics, proteomics, or drug discovery, and what outcomes they generate.

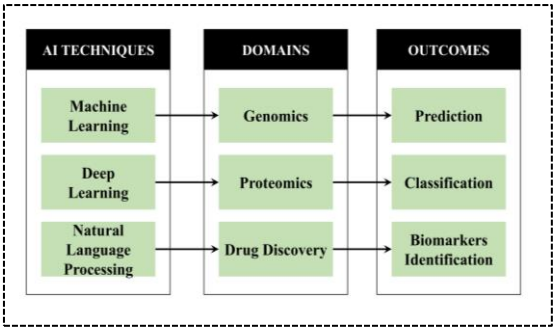

Fig. 2. Outcomes of AI-Bioinformatics Integration

A. AI and Omics

Omics describes a large scientific domain, such as genomics, metabolomics, and proteomics, each studying the large number of molecules found in living organisms. ML and DL are the two subfields of AI that are currently revolutionizing omics research. It helps to perform complicated data analysis and can help researchers explain various biological phenomena, diagnose diseases, as well as create individual treatment plans [45]. A study introduced a machine learning model based on eXtreme Gradient Boosting (XGBoost) library to classify renal tumors. This model is effective in distinguishing pathological subtypes in large-scale tissue datasets [46]. AI methods are essential for managing the high dimensionality of multi-omics data. Moreover, they play a pivotal role in uncovering hidden patterns, enhancing biological predictions, improving biomedical research, and facilitating data-driven decision making and advancement in personalized medicine [47].

B. AI and Genomics

It was found that DL models like Recurrent Neural Networks (RNNs) and Convolutional Neural Networks (CNNs) are quite efficient in sequence analysis as they have been used in Transcription Factor Binding Site Prediction (TFBSs), DNA motif discovery, and non-coding RNA element prediction. Likewise, CNNs split sequences, employ filters to examine parts, and define essential characteristics that establish labels, including DNA binding targets. Deep Variant uses CNNs for variant calling from the raw sequencing data, absolving it from these traditional problems like sample preparation, sequencing technology, and biological variability, and getting higher accuracy in variant calling [16]. CNNs are widely being used as a sequence classifier due to their ability to learn local spatial features from the dataset. A typical CNN contains a layered architecture containing an input layer, a convolution layer, an activation layer, and a pooling layer, followed by a dense layer [48]. The formulation of the convolution layer is as follows:

$$z_i = \int\left(\sum_{j=0}^{k-1} w_j . x_{i+j} + b\right) \quad (1)$$

where b in the equation represents the biasness, x is the input sequence, w is the size of the kernel, and f is the non-linear function. On the other hand, RNNs proceed through the character of the sequence by character and depend on the nearby context of a sequence to differentiate between the two areas, such as the exons and the introns in the DNA sequence [14]. More recently, researchers integrated data from different genomic platforms, including RNASeq, SNP, and CNV, with clinical follow-up data from TCGA. They used Random Forest (RF) analysis and found that the six-gene signature, made of CD24, PRRG1, MRGPRX, CASP8, RCC2, and IQSEC3, stands as one of the superior prognostic predictors for BC patients. These may help provide diagnostic and prognostic information, as well as provide the basis for the identification of targeted therapeutic pathways among the patient population [49].

C. AI and Transcriptomics

A comprehensive analysis of RNA transcription focusing on its structure, interaction, and functions in the expression of a specific gene. Integrating AI in this domain has significantly increased the prediction accuracy of complex patterns found in RNA sequences [50]. Deep learning models such as CNN and RNN are widely being used to address challenges such as complex pattern predictions, RNA sequence analysis, RNA and protein interaction, and analysis of non-coding RNA [51-52]. Takeshita proposed a machine learning-based framework for hormone receptor-positive breast cancer using logistic regression and hierarchical clustering, and predicted nine gene expressions, including C1orf64, AGL, CYP4F22, and others linked with the pathways of breast cancer of triple-negative value. Their proposed model efficiently predicts and highlights the tumor immune environments and pathways across different groups of patients. Further, the prediction accuracy was supported by chemotherapy and endocrine therapy [53-54].

D. AI and Epigenomics

The study of gene expression modification that occurs without changes in DNA sequence. AI enables researchers to handle large, complex data of sequencing produced from ChIP-seq and DNA methylation studies. CNNs are widely used to predict complex patterns in large sequences, including enhancers, transcription factor binding sites, promoter regions, and regulatory motifs. SVM has also been applied to predict disease risk at the individual level and facilitate early prevention strategies [55-56]. DNA methylation, histone-based modifications, human genome data, and RNA sequence data have been extracted using R programming for The Cancer Genome Atlas (TCGA). Nine different techniques were applied alongside eight different ML classification methods to determine the most precise model while focusing

on 1000 features across the data. To determine the most optimal model, k-fold cross-validation with k=5 has been applied and tested on different training and testing ratios by selecting 140 features using the Relief F feature selection method combined with the XGboost classification algorithm. The model achieved an accuracy of more than 90% and demonstrated an Area Under the Curve (AUC) for predicting liver cancer [57].

E. AI and Proteomics

The domain of proteomics mainly focuses on the structure, biological function, and protein interactions with DNA. AI is being used as a powerful adopted tool for unveiling the complex patterns present in macromolecules. Graph Neural Networks (GNNs) and CNNs are employed for the prediction of protein structure, protein-protein interaction, and drug development for targeted therapies and drug discovery [58-59]. Recent studies integrate machine learning models with microfluidic chips to analyze Extracellular Vesicles (EVs) in Triple-Negative Breast Cancer (TNBC). Researchers examined 100 breast cancer patients and 30 healthy individuals using microfluidic chips to isolate tumor-derived EVs to analyze EV proteomes. The machine learning model predicted distinct protein signatures differentiating TNBC patients from healthy individuals. This study opens up a new door by serving as a diagnostic tool and increasing recurrence in TNBC patients [60].

F. AI and Metagenomics

AI is transforming the field of metagenomics, which mainly focuses on analyzing the collective genomes of microbial communities. Machine learning algorithms, such as random forest and deep learning algorithms, address complex tasks like taxonomic classification and functional prediction [61]. Harris proposed a random forest model to identify large metagenomics datasets and achieved 91% accuracy in identifying the sample origin and predicting new samples while focusing on taxonomic profiles. This study highlights the ability of AI models to unveil hidden patterns of the microbiome and their effects on human health [50]. Random forest classification was proposed in a study to demonstrate improved accuracy in predicting colorectal cancer status in sequence data. Improving the predictive accuracy for colorectal cancer detection, AI-based novel approaches were used to integrate relative abundance profiles for both known and newly discovered microbes [62].

G. AI and CRISPR Data Analysis

CRISPR (clustered regularly interspaced short palindromic repeats) is a genome editing method extensively used in functional genetic research and has a significant impact on biomedical translation applications [15]. Integration and advancement in AI help enhance genome editing and improve precision, efficiency, and address diseases such as cell anemia. DeepCRISPR, CRISTA, and DeepHF are a few of the deep learning-based methods that are capable of predicting optimal guide RNAs for specific sequences. CRISPR and AI integration are revolutionizing healthcare by enhancing precise and personalized treatments. AI-based CRISPR system enables researchers to target genes that are pathways for disease. AlphaMissense is an ML model that predicts genetic variants for pathogenicity with high accuracy and surpasses traditional methods, and improves personalized medicine [63]. A study demonstrated that deep learning algorithms can be applied to identify rare diseases and their association with candidate genes. These CRISPR-based genetic modifications can be used to predict mutations and related genes. AI and CRISPR integration for genetic profiling can help analyze individual diseases, enabling timely interventions. These methods are delivering precise and reliable healthcare solutions with efficiency [64-65].

H. AI and System Biology

Network and system biology are the fields that provide a comprehensive understanding of biological systems by examining their structures, functions, and interactions. Data from multi-omics and its integration with AI enables researchers to model complex biological systems, focusing on valuable insights and relationships of genes, RNAs, proteins, and individual cells. AI algorithms, including ML and DL, are widely being used to handle the complex data of biological systems [66-67]. The objective of bioinformatics is the integration of diverse biological data, which is referred to as omics data. Despite notable advancements, AI methods encounter challenges in effectively managing this integration. Addressing these limitations remains the priority for future research while emphasizing the development of approaches to enhance the reliability and utilization of integration methods. Utilizing strict constraints on parameter and graph representation, with an ongoing effort, can be used to address these challenges while focusing on the integration and development of ML algorithms [45] [68]. A study shows the potential of ML algorithms when combined with system biology analysis in predicting cancer cell lines. Naïve Bayes and KNN algorithms show promising results while classifying cisplatin-resistant and sensitive samples. These findings provide the foundation for future experiments and development in this field [69]. AI transforms system biology by accelerating data analysis, prediction, and customized medicine. However, utilizing the full potential of AI models, several challenges, including data quantity, data-driven interpretation, ethical concerns, and privacy challenges related to data usage, may introduce biases in automation [70].

I. AI and Customized Medicine

The precision and customized medicine approach for medical treatment and proactive prevention strategies takes into account an individual's unique genetic information, environmental influences, and lifestyle. Artificial intelligence is advancing in every field day by day, and it is transforming the field of personalized medicine as well, by providing distinct opportunities for tailored medical care for individual patients [71]. Technologies based on AI, such as ML and DL algorithms, aid in finding and validating biomarkers by effectively analyzing extensive biological and clinical trial datasets, uncovering hidden patterns, and producing personalized treatment [72]. AI technologies, when combined with genomics and molecular biology, are improving customized medicine, making it more precise and reliable. This method focuses on finding diagnostic and hidden predictive biomarkers to enable patients to get custom-targeted medicines. [73]. To examine important gene markers in colorectal cancer, researchers used a variety of machine learning methods, such as logistic regression, support vector machines, decision trees, k-nearest neighbors, and random forest [58]. The random forest algorithm was the most successful for this prediction test because it showed the best accuracy and AUC. Differentially Expressed Genes (DEGs) and potential prognostic biomarkers in pancreatic cancer have been subjected to analysis using machine learning approaches such as RF, max voting, XGBoost, GBM, and AdaBoost. XGBoot surpasses all other models in processing speed and accuracy [74]. A study also reported that ML techniques, including SVM, random forest, logistic regression, and naïve bayes, are reliable models for biomedical data analysis [60]. High throughput screening has enhanced the detection of molecular interaction advancement in AI, but there still exist numerous possibilities and complexities that make targeted drug pairs and drug discovery a long and costly process [9]. For example, graph neural networks can predict potential drug molecules with targets while developing drugs. Generative adversarial networks increase the discovery process by generating novel drug molecules with the required properties. Moreover, AI models can forecast drug treatment efficiency, resulting in optimized target therapy and personalized medicines [75-76].

J. AI and Signal Interpretation

Signal processing involves the precise manipulation, comprehensive analysis, and interpretation of signals. Cardiac Electrocardiogram (ECG), Electroencephalogram (EEG), and Cardiac Electromyogram (EMG) are the types of physiological data signals that are being used for research and development. AI models can learn complex patterns in signal data and extract meaningful information while reducing noise and improving predictions in the health sector [77-78].

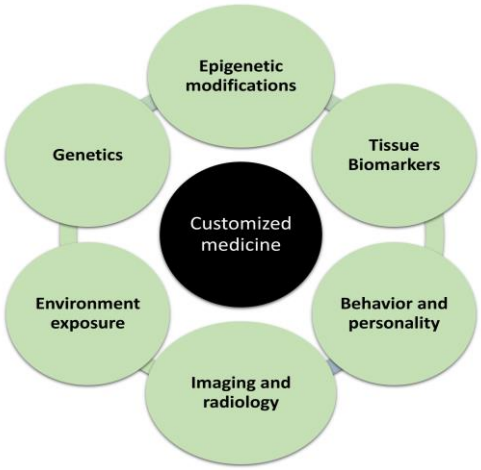

Fig. 2. Customized Medicine

The deep belief network has been utilized to analyze the EEG signals' frequency for the classification of motor images of the left and right hands. This study applies two AI models, SVM and DBN, and highlights that the DBN outperforms the SVM in all test cases. This study offers a deep learning method for correctly categorizing motor image-based EEG data [79]. A semi-supervised deep learning framework has also been proposed by researchers for recognizing states for EEG signals. This study outperforms traditional methods while handling EEG classification accuracy and improved adaptability, and performance [80].

K. AI in Biomedical Text Analysis

The vast literature of biomedical research provides a crucial knowledge base for researchers. To extract valuable insights and patterns from a variety of biomedical texts, including academic papers, clinical notes, and medical records, text mining approaches are being used. The pipeline combines two techniques, including natural language processing and machine learning for text translation and classification, respectively, alongside data mining techniques to identify underlying patterns and relationships. Named entity recognition (NER) is an NLP technique that is essential for extracting meaningful knowledge when accurately identifying entities such as gene sequences, protein sequences, and sequences of diseases. Categorizing and detecting named entities in the text while using the NER technique enables unmanned systems to accurately interpret and extract meaningful sequence patterns [81]. Several ML methods, such as SVM, Markov Models, and Maximum Entropy (ME), are widely being used for recognizing NER tasks [82-84]. Deep learning models such as CNN are extensively being used in biomedical text analysis. Application of CNN in the biomedical text is Transcription Factors Binding Sites (TFBSs)

classification, promoter analysis, telomeric analysis, and recurring pattern classification [85].

IV. Performance Evaluation

Numerous mathematical and evaluation metrics are employed to ensure the AI model's effectiveness and reliability when applied to the bioinformatics domain. To effectively evaluate the performance of a model, these metrics are crucial, especially in classification and regression tasks. Below are a few evaluation parameters that present the AI model performance [86-89]. The evaluation parameters are as follows:

$$Accuracy = \frac{TP+TN}{TP+TN+FP+FN} X\ 100 \quad (2)$$

$$\text{Sensitivity/Recall} = \frac{TP}{TP+FN} X\ 100 \quad (3)$$

$$\text{Precision} = \frac{TP}{TP+FP} X\ 100 \quad (4)$$

$$F1 - \text{Score} = 2\ x \frac{\text{Precison} * \text{Recall}}{\text{Precison} + \text{Recall}} X\ 100 \quad (5)$$

where TP stands for true positive, TN stands for true negative, FP stands for false positive, and FN stands for false negative. Loss functions such as Cross-entropy and mean squared error (MSE) are used for classification and regression to optimize the model's prediction performance.

Cross-entropy loss can be represented as:

$$L = -\sum_{i=1}^{n} y_i \log(\hat{y}i) \quad (6)$$

Mean squared error (MSE) can be represented as:

$$L = \frac{1}{n} \sum_{i=1}^{n} (y_i - \widehat{y_i})^2 \quad (7)$$

V. Discussion

Traditional bioinformatics methods are limited in handling complex and rapid biomedical data while relying only on pre-defined rules and a lot of human intervention. AI-driven approaches can process large and complex datasets, uncover hidden patterns, and improve prediction accuracy. Machine learning models can process large datasets, learn hidden patterns, and make accurate predictions. AI methods offer potential transformation in bioinformatics, such as personalized medicine, drug discovery, and identification of biological pathways [35] [39]. AI-driven approaches are promising in transforming bioinformatics data into valuable insights while overcoming the traditional methods in numerous fields of biomedical. However, to fully utilize the power of AI, a few challenges need to be addressed, such as data quality, model interpretability, and ethical considerations. Overcoming these challenges will help biomedical research and improve human health.

VI. Gap Analysis

Despite significant advancements, several research gaps and challenges exist in integrating AI into bioinformatics. AI models such as deep neural networks are considered black boxes and show high performance in prediction tasks. The lack of interpretability of these models restricts adoption in crucial medical situations. The development of AI models, such as explainable AI (XAI), ensures interpretability with high accuracy. Most AI models are data-specific with poor generalization. In the future, researchers must focus on the development of robust AI models with good generalization. AI models have yielded significant improvements in single-omics data, while multi-omics data integration is still emerging. The key challenges are to make these heterogeneous data feature selection and normalization. AI models rely on clinical and genetic datasets, making privacy and security crucial. To address these challenges, federated learning offers decentralized learning that makes personalized information secure. Transfer learning has also demonstrated promising results in bioinformatics, but its applications are still limited. These pre-trained models offer significant potential to increase predictive performance and reduce computational cost in domain-specific tasks.

VII. Conclusions

This review emphasized the pivotal role of AI in the field of bioinformatics, bridging biological data with computational methods. It covers a wide range of applications of AI techniques, including machine learning, deep learning, and natural language processing, and their impact on genome sequence analysis, protein folding and structure study, drug discovery, customized medicine, signal processing, and biomedical image analysis. The findings of this study show the contribution of AI in clinical and biomedical research, enhancing the performance in the prediction of pathways and personalized therapies. Transformers, Explainable AI (XAI), and multimodal learning are the promising domains of AI that offer robust performance, leading to innovation. The integration of these efficient and interpretable models encourages researchers to address complex problems in biomedical science.

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