




The Role of Artificial Intelligence in Advancements in Genetics: A Literature Review and Future Perspectives



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ABSTRACT

A combination of artificial intelligence (AI) and genetics has revolutionized biomedicine and pharmacology. It has opened new horizons toward analyzing complex genetic data and designing precise treatments. Significantly increasing amounts of genetic data and advancements in sequencing technologies have made AI a key tool for processing, analyzing, and identifying the patterns related to these data. The present work focused on the role of AI in the recent developments of genetics. It examined diverse applications of AI in diagnosis, prediction of therapy responsiveness, and improvement of gene editing technologies. In the realm of diagnosing genetic diseases, AI algorithms assist in the rapid and precise detection of rare diseases through analyzing genetic and clinical data. In addition, predicting responsiveness to particular treatments based on genetic data derived from predictive AI models may improve treatment outcomes and cut costs. AI also plays a critical role in designing and optimizing gene editing technologies, like CRISPR, and enhances the accuracy and efficiency of these methods. However, some challenges are important, like the quality of genetic data, the complexity of AI models, and ethical issues related to privacy and discrimination in using these technologies. The present work emphasizes the enormous potential of AI in genetics. It also pinpoints the importance of resolving challenges for more effective and equitable utilization of this technology.

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Introduction

A

Necessity and importance

combination of artificial intelligence (AI) and genetics has revolutionized biomedicine and pharmacology. Advances in genomic techniques and the availability of extensive genetic data allow for significantly broader application of AI algorithms to analyze these data. This combination may assist in identifying more complex patterns within genetic data, leading to the development of more accurate therapeutic methods. For example, using AI, researchers can detect genetic factors related to diseases, predict responses to treatments, and improve gene editing methods [1-4]. Moreover, given the vast amounts of available data and their growth rate, it is difficult to analyze them manually, and AI provides the necessary tools to overcome these challenges [5, 6].

Goals

The present review focused on the role of AI in recent advancements in genetics. The following were discussed:

How to analyze genetic data and identify complex patterns using AI.

Diagnosing and treating genetic diseases with AI.

The current challenges and obstacles of using AI and the future perspectives.

Basics

AI is a set of algorithms and techniques capable of simulating human tasks, such as learning, reasoning, and problem-solving [7]. Genetics studies genes, inheritance, and genetic diversity of organisms. Genes are inherited units carrying the genetic materials required to determine the physical and biochemical characteristics of living beings [8-10]. Changes in these genes may lead to disease or affect treatment responsiveness. As a recent advancement in genetics, gene editing allows for the modification of genetic information, and AI can assist in improving this process [11, 12].

Applications of AI in genetics

AI is a powerful tool for studying genes and improving human health (Figure 1). Some of these applications are explained below.

Genetic data analysis

Genetic data are among the largest and most complex data resources in biological sciences. AI algorithms allow us to analyze these data rapidly and accurately [13-15]. For example, AI helps us analyze next-generation sequencing (NGS) data. It can also be effective in detecting alterations in DNA sequences and interpreting their biological outcomes [16-18]. Algorithms, like artificial neural networks and support vector machines, are particularly applicable in identifying nonlinear and complex patterns in genetic data. These can help us predict complications and symptoms of the diseases [2, 19, 20].

Disease diagnosis

AI can also be used to diagnose genetic diseases early and accurately. Recent studies using AI models to analyze clinical and genetic data suggest that these algorithms can aid in the more rapid and accurate detection of rare diseases. These approaches not only increase diagnostic accuracy but also reduce the time required for diagnosis [21-23].

Prediction of treatment responsiveness

Predictive models using AI can help physicians predict patients' responses to specific therapies based on their genetic makeup. Using genetic and clinical data, AI algorithms assist in identifying patterns related to patients' responses to particular treatments. For example, investigators have demonstrated that using AI, treatment outcomes might be predicted in patients with cancer. In this way, physicians can concentrate on more effective treatments and avoid ineffective ones [2, 24-28].

Gene editing and developing gene editors

Gene editing techniques, like CRISPR (clustered regulatory interspersed short palindromic repeats), have become one of the most innovative tools in genetics, and AI can assist in designing and improving these technologies [29]. AI may contribute to identifying proper target sites for gene editing and predicting the potential side effects. For instance, deep learning algorithms may help in predicting the potential effects of gene editing on gene function and cellular circumstances [8, 30, 31]. By analyzing big data such as sequencing data or data related to protein structure, AI techniques provide the potential for optimizing the gene editing process. In this way, the accuracy of these techniques increases significantly [32, 33].

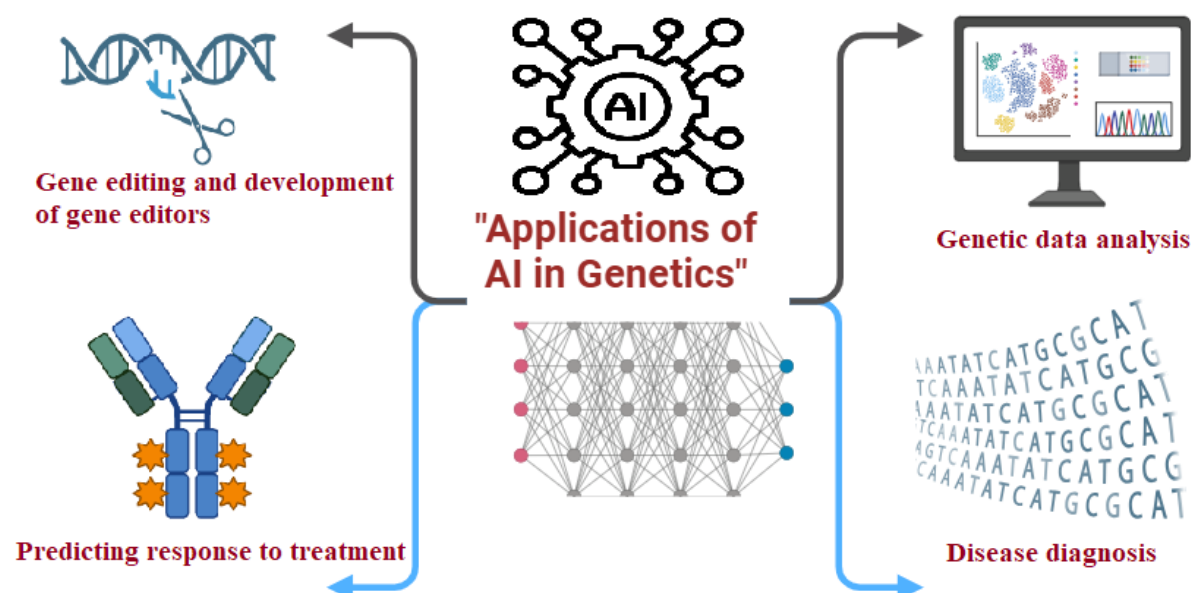


Figure 1. Various applications of AI in genetics



Challenges and obstacles

Technical challenges

The application of AI in genetics presents several technical challenges. One of the biggest challenges is data quality. Genetic data are highly variable and extensive, often including high error rates. For instance, NGS techniques may suffer from rare errors that can affect their final results [34, 35].

Data interpretation is another challenge. AI algorithms can present accurate results. However, a lack of clarity on how to gain these results may be challenging for researchers and medical specialists. This lack of clarity can make the data interpretation difficult, affecting the reliability of the results [36, 37].

Ethical issues

With the increasing application of genetic data in AI algorithms, there is a risk of leaking critical information, which may lead to potential discrimination and abuses. Ensuring the protection of genetic data and its appropriate encoding is one of the most critical challenges. One of the biggest concerns is genetic privacy [38].

Besides privacy, issues related to discrimination are also among the important ethical aspects. If AI algorithms are trained on false or non-representative data, it may lead to discriminatory results [39].

Ultimately, issues related to social and economic vulnerability in the application of AI in genetics are also important. Ensuring equitable access to new technologies and preventing health gaps among different demographic groups poses a serious challenge for researchers and policymakers [40, 41].

Future perspective

Future innovations

Given the broad and rapid advances in AI and genetics, we expect to have key innovations in combining these two fields. One of the innovative areas is the improved accuracy and efficacy of deep learning algorithms for analyzing genetic data. Using extensive data and new techniques, like reinforcement learning, we can develop more accurate and general models [40, 42].

It is expected that the advances in sequencing technologies will provide genetic data with higher quality and quantity [43]. The application of AI in this multidimensional space may assist in the identification of more complex patterns occurring in physiological and clinical data. Still, developing more understandable and clearer algorithms remains a requirement for public trust building and acceptability [44].

Finally, it is critical to note the combination of new techniques in the realm of genetics, such as integrating gene editing with two methods—CRISPR and ACT-G

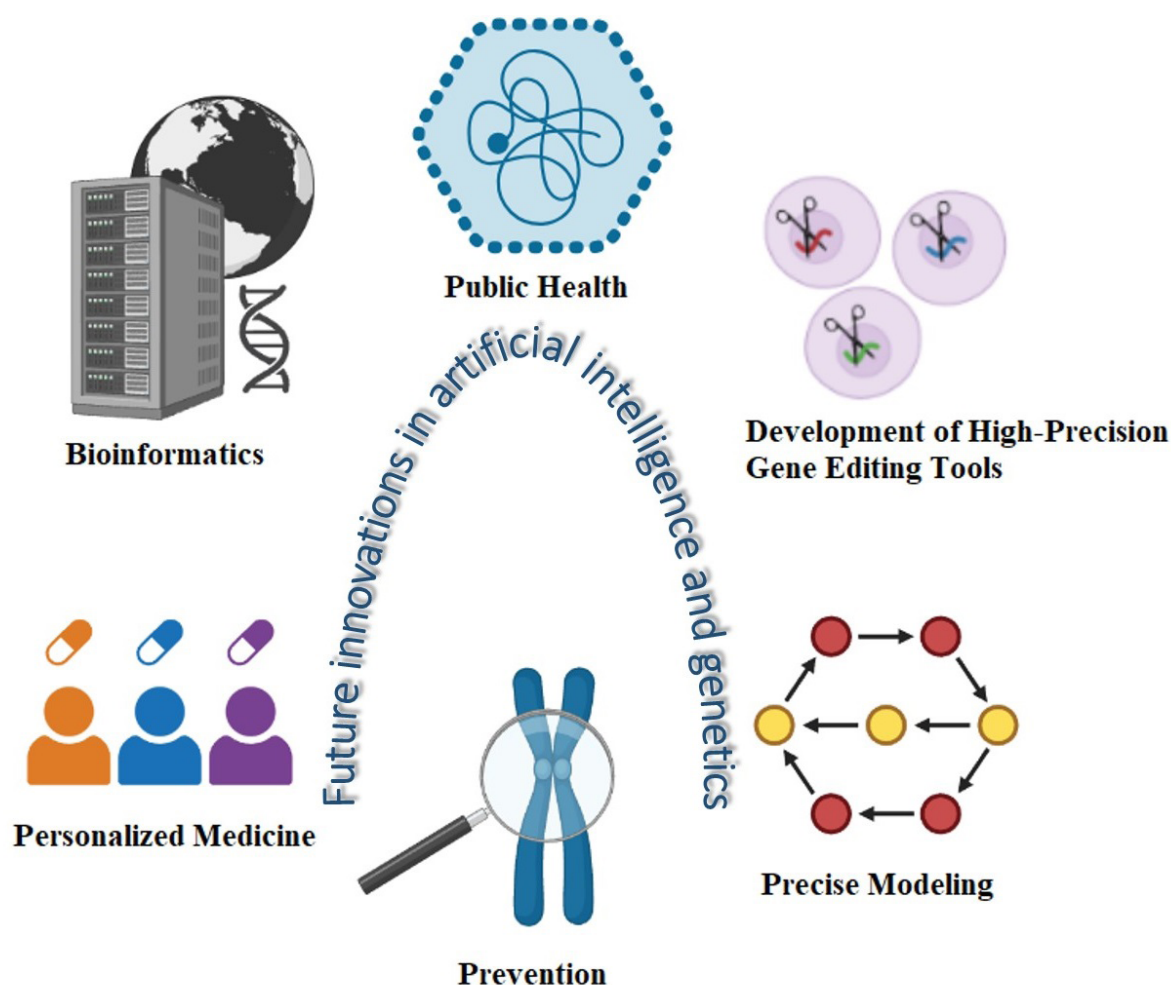


Figure 2. Emerging trends in combining AI and genetics



(acceptance and commitment therapy)—with the aid of AI for more accurate design and optimization. This integration means the availability of better and more accurate results in gene editing and reduced potential side effects [45, 46].

Emerging trends

There are several emerging trends in combining AI and genetics, which may have potentially profound effects on medicine and therapy (Figure 2).

Bioinformatics and big data: Over time, the amount of genetic and clinical data has increased significantly. Exploiting bioinformatics technologies combined with AI may enable the processing, analysis, and interpretation of big data. This trend helps researchers in the realms of drug discovery, identification of disease-causing genes, and analysis of biomarker variables [47, 48].

Accurate modeling and personalized medicine: One of the most important future trends is the personalization of treatments based on an individual's genetic data. Using AI, it is possible to design personalized therapeutic profiles for patients, allowing therapies to be optimized based on each individual's genome [49-51].

Developing high-accuracy gene editing tools: The recent advances in gene editing technologies, like CRISPR-Cas9 and upgrading to newer methods, such as CRISPR-Cas12, allow scholars to use AI for designing and implementing gene editing with better outputs. It has significantly increased the accuracy and effectiveness of these methods [52-54].

Public health and disease prevention: The use of AI to identify genetic risk patterns and diseases at the population level can aid in disease prevention. This trend includes the analysis of clinical and genetic data to detect individuals at risk and suggest preventive solutions [55, 56].

Conclusion

AI and its capabilities in analyzing complex and vast data play an irreplaceable role in the development of genetics. From identifying complex patterns in genetic data to advances in diagnostic methods, predicting treatment responsiveness, and developing gene editing techniques, AI presents new capabilities for treatment and research. This may set the stage for the introduction of new therapies for common diseases in the future. Despite technical and ethical challenges, using this technology in conjunction with new genetic techniques promises a bright future with further advancements. More investigations in this realm may contribute to more accuracy, efficacy, and reliability in AI applications.

Ethical Considerations

Compliance with ethical guidelines

This article is a review article with no human or animal sample.

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Authors contribution's

Conceptualization, resources, project administration and supervision: Seyed Hossein Khaleghinejad; Methodology: Yousef Sefidi Heris and Dariush Gholami; Software and writing the original draft: Yousef Sefidi Heris; Validation and investigation: Dariush Gholami; Formal analysis: Seyed Hossein Khaleghinejad and Yousef Sefidi Heris; Review and editing, Seyed Hossein Khaleghinejad and Dariush Gholami.

Conflict of interest

The authors declared no conflict of interest.

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