

Artificial Intelligence in Genomic Research and Advanced Healthcare Applications

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ABSTRACT

Genomics has emerged as a cornerstone of modern biomedical research, enabling the comprehensive study of genome structure, function, and variation across organisms. With the advent of high-throughput sequencing technologies and the accumulation of large-scale omics data, AI, ML, and DL have become indispensable in complex genomic analysis. The integration of AI across the broad spectrum of genomic disciplines, including structural and functional genomics, and specialized subfields such as epigenomics, pharmacogenomics, metagenomics, and cancer genomics, is explained. In structural genomics, AI models have accelerated disease diagnosis and drug discovery, predicting gene mutations and mapping genome architecture. Genomics powers AI, biomarker discovery, microarray analytics, and genome editing. AI facilitates predictive medicine, disease surveillance, streamlining treatment protocols, and augmenting patient care. The integration of AI in genomics is enhancing diagnostic precision and therapeutic strategies, shaping the future of personalized and population-scale healthcare.

INTRODUCTION

The study of an organism's entire genome is termed Genomics, and it has rapidly evolved as a transformative field at the intersection of biology, data science, and medicine, further enabling insights into gene structure, function, and their interaction (Lesk, 2017; Del Giacco & Cattaneo, 2011). Advances in sequencing technologies and computational tools have helped genomics researchers beyond basic DNA mapping. Current research includes functional, structural, and comparative studies across different or-

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ganisms (Hocquette, 2005; Hardison, 2003). With the availability of large genomic datasets, researchers can explore complex genetic structures, identify how diseases develop, and contribute to the growth of personalized healthcare (Pevsner, 2015; Bustamante et al., 2011). The growing attention towards medical genomics and studies on specific populations highlights its importance in precision medicine (Seehausen et al., 2014; Cooper, 2003). Precision medicine is the utilization of modern tools to create specified medication and treatment protocols that consist of optimal actions for the patient without any side effects, based on available data. Precision medicine aids in improving health outcomes by early diagnosis and better prognosis for chronic diseases. It relies on the clinical data associated with the patient, as well as their genomics and multi-omics (transcriptomics, proteomics, metabolomics, phenomics, etc.) data, to determine how a physician should continue to recommend a specific treatment regimen.

Artificial Intelligence (AI) is a general term that elucidates the process of using computers and digital technology to create thought-provoking software that resembles human-like critical thinking. It simulates human cognitive functions such as learning, reasoning, and decision-making by machines. The subfields like machine learning (ML) and deep learning play a central role in data-driven artificial intelligence tasks (Hunter, 1993; Gao et al., 2022). Genomics research generates vast amounts of complex data, and AI has emerged as a transformative tool for analysing, interpreting, and extracting meaningful insights from this information (Boulesteix & Wright, 2022; Guo et al., 2023). In particular, AI facilitates the annotation of genomes, identification of disease-associated variants, and prediction of gene-disease relationships with speed and accuracy than traditional methods (Dias & Torkamani, 2019; Xu et al., 2019). Deep learning architectures are enabling precision medicine by integrating multi-omics data to uncover hidden patterns in genetic disorders (Quazi, 2022; Ahmed et al., 2023). While AI has significantly advanced human health research, it has also proven valuable in plant and microbial genomics, highlighting its wide range of applications across various biological fields (Maqsood et al., 2024; D'Agaro, 2018). The AI and genomics are enhancing diagnostic capabilities, accelerating drug discovery, and the development of personalized therapies (Álvarez-Machancoses et al., 2020; Williams et al., 2018).

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