REVIEW ARTICLE

# The Role of Artificial Intelligence (AI) in Genomics

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## Abstract

Artificial Intelligence (AI) is creating computer systems capable of performing tasks that require human intelligence. Medical AI applications have recently attracted significant attention due to advancements in AI hardware and software, deep learning algorithms, and graphics processing units (GPUs) that enable their training. While other AI subtypes have started to show similar progress in multiple diagnostic modalities, AI-based computer vision methods are poised to transform image-based diagnostics into clinical diagnostics. In specific fields, such as therapeutic genomics, large and complex genetic data are processed using a particular type of AI algorithm called deep learning. Numerous practical applications across a range of industries are being transformed by artificial intelligence (AI). Several conventional machine-learning techniques have been applied in genomics to comprehend the dynamics of genetic data. AI enables rapid identification of genetic variants, determination of gene functions, and the uncovering of intricate genetic relationships by combining machine learning algorithms, deep learning models, and data-driven insights. AI-powered technologies have significantly cut costs and time by streamlining the genome sequencing, annotation, and editing procedures. Additionally, AI supports personalized medicine by evaluating each person's unique genomic profile to forecast illness risks, enhance treatment regimens, and find new therapeutic targets. Additionally, the combination of AI and genomics has accelerated advances in agricultural genomics, drug development, and biological evolution research.

**Keywords:** *Artificial Intelligence (AI), genomics, machine learning, deep learning, medical AI applications, genome sequencing, therapeutic targets, drug discovery.*

## INTRODUCTION

The fundamental tenet of molecular biology, put forward by Francis Crick in 1957, describes how genetic information moves from DNA (deoxyribonucleic acid) to RNA (ribonucleic acid) and then from RNA to protein (a functional form of DNA) in a living organism [1]. Four fundamental building blocks known as nucleotides, adenine (A), thymine (T), cytosine (C), and guanine (G), are found in the double-helical strands of DNA. A, T, C, and G are joined together, forming a chemical bond between the two strands of DNA. Everything that is biological and needs to be translated into a protein output is contained in the DNA base sequence. DNA is arranged in living cells as chromosomes, which are further subdivided into segments of DNA known as genes that code for proteins [2]. The genome is an organism's complete set of genes. The study of a genomic sequence or an organism's genome is called genomics, a branch of science [3]. Approximately 3 billion DNA base pairs comprise the human genome, and the study of human genomics aims to associate the genome with biological and physical characteristics. Next-generation sequencing (NGS) technology is a component of this data-driven study, which yields information about an organism's entire genome. Whole-genome sequencing (WGS), exon sequencing (WES), chromatin and epigenetic profiling, and transcriptomics are among these sequencing techniques [4]. The Human Genome Project (HGP), which provided the framework for most of the human genome, concluded in 2001, marking an essential technical development in genomics [5]. Thanks to recent advances in long-read technology, the remaining 8% of the human genome has been sequenced, illuminating the human genome reference.

Genetic variation among animals, or even within an organism's many cells, organs, and disease states, has been enhanced by whole-genome sequencing. The consequences of the Human Genome Project (HGP) showed that 99.9% of human beings are heritably alike and that only 0.01% of the human genome can trigger phenotypic alterations in people, comprising drug responses, disease vulnerability, and physical characteristics (*e.g.* intelligence, height, eye color, *etc*.). Identifying the underlying alterations or mutations in DNA sequences that can modify cellular functions and lead to disease states is a primary goal of genomics. This is typically accomplished using genome-wide association studies (GWAS) [6]. It is important to remember that not all mutations that occur cause disease. For instance, not all indels (insertions or deletions of small pieces of DNA) or single-nucleotide polymorphisms (SNPs) alter the DNA sequence that codes for a protein (synonymous mutations) or the expression of the genes [7]. We will be able to develop better medications, therapies, or even cures if we can determine which variant is associated with a particular illness.

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*Received: March 24, 2025; Revised: August 16, 2025; Accepted: August 19, 2025*
*DOI: https://doi.org/10.37184/jlnh.2959-1805.3.50*

  

One of the most common chronic illnesses brought on by genetic modification is cancer, including amplifications, base changes, deletions, and rearrangements. Various forms of cancer have multiple pathways for sequence change [8]. Additionally, genomics is crucial to the study and management of infectious illnesses at the individual and population levels. It specifically aids researchers in detecting and monitoring the development of drug resistance in pathogenic organisms. For instance, during the COVID-19 pandemic, genomics assisted researchers in tracking the spread of the virus (pathogen) and gaining insight into how the strain was changing to help create effective vaccinations [9]. More focused tests, such as those for rare diseases, tumor genome sequencing, and non-invasive prenatal screening, are also made possible by genomics.

Additionally, by helping researchers understand the genetic composition of crops and cattle, genomics has transformed the agricultural industry. Using genomics, scientists will be able to generate genetically modified organisms (GMOs) that are more resistant to pests, tolerant of severe ecological conditions, and capable of producing more. It is obligatory to address the problems posed by the world's growing population and ensure food safety. Associating the genomes of diverse species and investigating the significant concepts of organismal environmental adaptations and evolutionary histories can also help us recognize biodiversity [10].

## AI IN CANCER GENOMICS

The progression of cancer is triggered by alterations or mutations in the DNA sequences of genes that regulate cell division, survival, or other features of different phenotypes. This leads to uncontrolled cell growth and the spread of abnormal cells [11]—genetic abnormalities that impact normal cell development pathways and cause tumor formation can be acquired by these cells. Researchers have been attempting to understand the fundamental underpinnings of tumors with varying clinical outcomes for several years. Understanding the fundamentals of this diverse and intricate illness can be gained through genomics [12]. Researchers can identify several altered genes, known as oncogenes, that cause cancer using genomic studies. The TP53 gene, which is changed in several malignancies, is a typical example [13].

We can find and comprehend clinically significant genetic variants that may be targeted for prospective therapeutics by using NGS technologies to profile a cancer patient's entire genome. In 1998, trastuzumab, the first molecular targeted medication based on comprehensive genomic profiling (CGP), was made available to patients with breast cancer that overexpressed ERBB2 [14].

Inhibitors of BRAF melanoma, BCR/ABL chronic myeloid leukemia, and epidermal growth factor receptor (EGFR) non-small cell lung cancer tyrosine kinases are among the new targeted medicines discovered since then and have shown promising results. The majority of large-scale genome initiatives, such as The Cancer Genome Atlas (TCGA) and the International Cancer Genome Consortium (ICGC), have focused on characterizing the cancer genome [15]. However, the human genome is 98% non-coding and only 2% coding, and little is known about how changes in the non-coding portion of the genome may influence the onset of cancer.

According to recent research, there is a strong correlation between disease conditions and changes in the non-coding regulatory region of the human genome [16]. In addition to being crucial for understanding how the genome functions, the identification and characterization of gene regulatory regions also help expand our knowledge of disease causation and provide a more comprehensive picture of disease status. For instance, one of the leading causes of brain cancer, glioblastoma, is a mutation in the regulatory region of the RB1 gene. Promoters, activators, enhancers, and silencers are among the various non-coding regulatory regions of genes [17]. This article will focus on accompaniment areas and identifying and understanding accompaniment mechanisms in cancer.

## AI IN GENOMIC SEQUENCING

Genomic sequencing has been developed using artificial intelligence (AI), which has enhanced interpretation, data analysis, and the understanding of complex genetic information [18]. AI speeds up the analysis of massive genomic datasets, increasing accuracy and reducing sequencing time by detecting errors and decoding intricate sequences [19]. The usage of annotation tools is crucial for identifying genetic variations such as insertions, deletions, single-nucleotide polymorphisms (SNPs), and structural aberrations, and linking them to diseases. AI assists with initial identification and analysis in personalized medicine by detecting disease-associated mutations, particularly in cancer genomics, where tumor-specific mutations drive personalized therapies [20]. Additionally, by examining gene expression and epigenetic changes, AI-powered methods enhance functional genomics by providing insights into gene regulation and activity [21].

By identifying therapeutic targets and developing medications tailored to genetic profiles, AI also supports drug discovery. AI is helping researchers better understand rare diseases by prioritizing candidate genes and integrating multi-omics data for comprehensive analysis. AI supports precision health efforts in population genomics by analyzing genetic variation and identifying disease risk factors unique to a group [22, 23].

## AI IN DISEASE DIAGNOSIS

Artificial intelligence (AI) is transforming disease diagnosis by improving the efficiency, speed, and accuracy of medical condition identification [24]. Large volumes of medical data, including imaging, genomic, and clinical records, are analyzed by AI-powered systems to identify trends and abnormalities that may indicate illnesses. In activities such as imaging analysis, machine learning algorithms often outperform humans and excel at identifying early indicators of diseases like cancer, heart disease, and neurological disorders. AI systems, for example, may detect cancers, fractures, and other anomalies with high precision in medical images such as X-rays, MRIs, and CT scans, helping radiologists make more precise diagnoses [25]. Artificial intelligence is crucial for the analysis and imaging of genomic data in the diagnosis of rare and congenital illnesses. AI marks early recognition, and personalized management plans are likely to result from distinguishing disease-related mutations. By patient history, symptoms, and lifestyle, AI-powered investigative platforms may also examine electronic health records (EHRs) and estimate the possibility of certain illnesses [26]. Using artificial intelligence (AI), epidemiological data analysis, and genetic sequencing helps manage infectious diseases by detecting infections and predicting epidemics.

A subfield of artificial intelligence (AI), natural language processing (NLP), enables the extraction of meaningful information from unstructured medical data, including study articles, which supports the development of manufacturing analyses [27]. Effective aides and Artificial Intelligence (AI) chatbots are also used to conduct early assessments, present patients with personalized recommendations, and refer them to the appropriate treatment. To ensure the safe and fair use of Artificial Intelligence (AI) in disease identification, issues such as algorithmic bias, data privacy, and the conditions for monitoring authorization must be addressed, despite these improvements. AI has the potential to transform patient care and improve diagnosis precision in healthcare with additional advances [28].

## AI IN DRUG DEVELOPMENT

Drug expansion is being developed using artificial intelligence (AI), which speeds up timelines, reduces costs, and updates processes [29]. It frequently takes more than 10 years to bring a medicine from concept to market due to the drawn-out, resource-intensive nature of traditional drug discovery. By evaluating large datasets, forecasting drug-target interactions, and identifying viable drug targets with previously unheard-of speed and precision, artificial intelligence (AI) addresses these problems [30].

To identify biological targets linked to diseases, machine learning algorithms examine genomic, proteomic, and clinical data. This is one of the primary uses of AI in drug development. AI-driven drug screening algorithms significantly reduce the requirement for costly laboratory testing by evaluating millions of chemical compounds to predict their potential efficacy and safety after a target has been discovered. AI is also essential for improving the drug design process, using deep learning methods to generate new compounds tailored to specific biological targets [31].

By selecting ideal patient demographics, predicting trial outcomes, and monitoring adverse events in real time, AI also improves the efficiency of clinical trials [32]. Identifying biomarkers that stratify patients can guarantee more individualized and efficient therapies. Insights from scientific publications and patent databases are also extracted using natural language processing (NLP) techniques, helping researchers stay current and identify new treatment options.

To ensure drug candidates have appropriate absorption, distribution, metabolism, and excretion (ADME) profiles, artificial intelligence (AI) helps predict pharmacokinetics and pharmacodynamics in later phases of research [33]. Additionally, AI algorithms assess potential toxicity, reducing the likelihood that preclinical or clinical trials will fail [34]. Furthermore, development timeframes are significantly shortened by the growing usage of AI-driven platforms to repurpose current medications for new purposes.

Even as AI has the potential to revolutionize drug research, there are still obstacles to overcome, including the availability and quality of training data, legal restrictions, and the need for collaboration between pharmaceutical scientists and AI specialists. However, with further developments, AI has the potential to completely transform drug development and deliver safer, more efficient treatments to patients more quickly than ever before [35].

## AI IN MEDICAL CARE

By increasing patient outcomes, optimizing workflows, enabling individualized therapy, and boosting diagnostics, artificial intelligence (AI) is transforming healthcare. AI helps healthcare providers make quicker, more accurate decisions by identifying patterns and insights from massive volumes of data [36]. It is increasingly used across many facets of healthcare, changing how illnesses are identified, treated, and managed.

In diagnostics, artificial intelligence (AI)-powered technologies evaluate medical imaging, including MRIs, CT scans, and X-rays, to identify anomalies like tumors, fractures, and disease indicators, frequently with accuracy on par with or better than human professionals' models, for instance, can detect malignancies or retinal disorders early on, allowing for prompt treatment. AI also improves accuracy in laboratory medicine, for example, by detecting infections in blood samples or identifying biomarkers associated with specific diseases [37].

By evaluating patient, proteomic, and genetic data to tailor treatment regimens to specific requirements, AI helps personalize medicine [38]. It assists physicians in choosing the best treatments with the fewest possible adverse effects by predicting how patients will respond to specific therapies. Wearable technology and AI-powered monitoring devices measure vital signs to support the management of chronic diseases, providing real-time insights into conditions such as heart disease, diabetes, and hypertension. These systems' alerts enable prompt action, which lessens difficulties.

AI also improves operational efficiency in healthcare facilities by automating administrative tasks such as billing, scheduling, and maintenance of electronic health records (EHRs) [39]. Natural language processing (NLP) helps doctors make decisions and lessen their workload by extracting insightful information from patient histories and clinical notes. By providing patients with trustworthy information, reminding them of their prescription regimens, and conducting initial evaluations, chatbots and virtual health aides increase patient access and participation.

AI has also helped telemedicine by facilitating remote consultations using sophisticated diagnostic instruments [40]. AI helps doctors diagnose and treat patients by analyzing their medical histories and symptoms during consultations. Furthermore, AI enables faster innovation in treatment and epidemic prediction, making it essential for drug research, vaccine development, and population health management [41].

Although its innovative influence is significant, issues such as data security, algorithmic bias, ethical dilemmas, and integrating AI tools into existing procedures remain essential. However, as technology improves, artificial intelligence (AI) has significant potential to improve healthcare efficiency, patient-centeredness, and affordability, opening the door to an emerging, data-driven approach.

## AI IN PERSONALIZED MEDICINE

### Genomic Analysis and Precision Treatments

Artificial Intelligence (AI) develops methods for analyzing genomic data to identify biomarkers, genetic variants, and alterations associated with specific diseases [42]. Machine learning algorithms can estimate how an individual's genetic configuration affects their response to drugs, enabling personalized treatment that maximizes efficacy and reduces adverse effects.

### Patient Stratification

Artificial Intelligence (AI) distributes patients into groups based on genetic, environmental, and clinical data using sophisticated techniques [43]. Patients with similar summaries are more likely to receive medicines that are most likely to help them, which improves outcomes and reduces the need for trial-and-error when making script recommendations.

### Predictive Modeling for Disease Risk

Artificial Intelligence (AI)-powered analytical representations estimate a person's genetic, medical history, and lifestyle evidence to define how likely they are to contract a specific disease [44].

### Multi-Omics Integration

Data from numerous omics layers of proteomics, transcriptomics, Metabolomics, and genomics are incorporated through AI to offer a systematic representation of a patient's biology [45]. This inclusive technique progresses personalized treatment plans by revealing complicated associations among genes, proteins, and metabolites.

### Personalized Monitoring and Feedback

Artificial Intelligence (AI) systems constantly monitor a patient's vital signs, including responses to therapy and lifestyle choices [46]. By continuously amending management plans based on real-time data, these organizations ensure patients receive the best care over time.

### Reducing Adverse Drug Reactions

Artificial Intelligence (AI) estimates how a patient's genetic makeup may affect adverse medication interactions. As of this skill, doctors can recommend harmless, patient-specific substitutes to unsafe medications [47].

### Population Health Insight

Artificial Intelligence (AI) uses population-level genomic data to discover patterns and insights that inform public health interventions and ensure that personalized care reaches more people, particularly underserved groups [48].

### Applications of AI in Genomics

Table **1** shows applications of AI in Genomics [18].

**Table (1):** Applications of AI.

| AI Application | Purpose |
|---|---|
| Genomic Sequencing | Rapid, precise analysis of genetic data. |
| Cancer Genomics | Rapid, precise examination of cancer-related genetic data. |
| Drug Development | Fast-tracking and improving drug discovery. |
| Medical care | Improving diagnosis and treatment. |
| Personalized Medicine | Customizing therapies for each patient. |
| Disease Diagnosis | Rapid disease identification |

The healthcare industry has seen a fundamental alteration as a result of the assimilation of Artificial Intelligence (AI) into genomics, disease diagnostics, medical treatment, medication discovery, and customized medicine, AI's capability to estimate massive volumes of genomic data, spot trends, and predicted consequences has sped up drug discovery, improved disease diagnostics, and permit for more personalized management procedures. Our data on multifaceted ailments has been developed using AI in genomics, including patient stratification, genetic analysis, multi-omics integration, and predictive modeling. Moreover, AI-powered tools have developed disease diagnostics, enabling early recognition and therapy. AI has also accelerated cost-cutting, drug development, and timelines [49]. Artificial Intelligence (AI) procedures have also enhanced patient outcomes, improved the quality of medical treatment, and enabled more efficient processes [50]. Nevertheless, issues such as data privacy, legal frameworks, and algorithmic bias persist. To certify the safe and effective integration of AI in healthcare, these issues must be addressed [51, 52].

## CONCLUSION

In conclusion, healthcare could be transformed by the integration of genetics and AI, enabling more personalized, precise, and effective treatments. We may expect remarkable revolutions in drug development, healthcare, and disease detection as Artificial Intelligence (AI) knowledge continues to advance, eventually improving human health and well-being.


## FUNDING

None.


## CONFLICT OF INTEREST

The authors declare that they have no conflict of interest.


## ACKNOWLEDGEMENTS

Declared none.


## AUTHORS' CONTRIBUTION

Safia Gul: Study design, data collection and analysis, and manuscript writing.

Ainul Hayat: Assisting in proofreading.

Saba: Conceived and designed the research idea.

Maryam Maqsood: Supervised the project, ensured the study's integrity, and handled the submission and revisions process.

## GENERATIVE AI AND AI-ASSISTED TECHNOLOGIES IN THE WRITING PROCESS

During the preparation of this work the author(s) limitedly used ChatGPT (GPT-4, OpenAI) to get language suggestions and do minor proofreading in some parts of the manuscript. After using this tool/ service, the author(s) reviewed and edited the content as needed and take(s) full responsibility for the content of the published article.

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