



UNLOCKING GENETIC INSIGHTS: EXPLORING THE POTENTIAL OF AI SYSTEMS IN GENE ANALYSIS

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ABSTRACT:

Gene analysis is a fundamental component of biomedical research and clinical practice, providing insights into the genetic factors influencing diseases, treatment responses, and individual variations. In recent years, the integration of artificial intelligence (AI) systems has revolutionized the field of gene analysis, enabling researchers and clinicians to unlock the vast potential hidden within genetic data. AI algorithms excel at processing large volumes of genetic information, extracting meaningful patterns, and providing accurate predictions, thereby accelerating discoveries and advancements in personalized medicine. This article provides an overview of the application of AI systems in gene analysis, highlighting their advantages, challenges, and future prospects. It explores the role of AI in data analysis, diagnostic support, and the development of predictive models for genetic diseases. Additionally, it discusses the importance of interpretability, ethical considerations, and the integration of AI with other technologies. As AI continues to evolve, it is poised to reshape the landscape of gene analysis, leading to more precise diagnoses, personalized treatments, and a deeper understanding of the complexities of the human genome.

Keywords: gene analysis, artificial intelligence, precision medicine, feature extraction

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INTRODUCTION:

Gene analysis, also known as genetic analysis, is a field of study that involves examining an organism's DNA (deoxyribonucleic acid) to gain insights into its genetic makeup and understand how genes contribute to various traits, diseases, and biological processes. It

encompasses a wide range of techniques and methodologies aimed at deciphering the information encoded within an individual's genes. The human genome is composed of billions of nucleotides arranged in a specific sequence, and each nucleotide can be one of four types: adenine (A), thymine (T), cytosine



(C), or guanine (G). Genes are segments of DNA that contain instructions for building and maintaining an organism. They determine various characteristics, including physical traits, susceptibility to diseases, and responses to environmental factors.¹Gene analysis involves several key steps. The first step is DNA extraction, which involves isolating DNA from cells or tissues. This can be done through various methods, such as collecting a blood sample, swabbing the inside of the cheek, or extracting DNA from other bodily fluids or tissues. Once the DNA is extracted, it undergoes a process called DNA sequencing, which determines the order of nucleotides in the DNA molecule.² There are different sequencing techniques available, with advances in technology leading to faster and more cost-effective methods. DNA sequencing generates large amounts of data, which can be challenging to analyze and interpret without computational tools. Bioinformatics plays a crucial role in gene analysis. It involves the use of computer algorithms and databases to store, retrieve, and analyze biological data, including DNA sequences. Bioinformatics tools help in comparing DNA sequences to reference genomes, identifying genetic variations, and predicting the functions of genes.³Genetic variations, such as single nucleotide polymorphisms (SNPs) or structural variations, can have significant implications for an individual's health and traits. Gene analysis techniques, such as genotyping and DNA microarrays, are used to detect and analyze these variations. Genotyping involves determining the presence or absence of specific genetic markers or mutations, while DNA microarrays allow the simultaneous analysis of thousands of genetic markers.⁴ Gene expression analysis is another important aspect of gene analysis. It focuses on understanding which genes are active and producing proteins in a particular tissue or under specific conditions. Techniques like RNA sequencing (RNA-seq) and microarray analysis enable researchers to measure the levels of gene expression and identify differentially expressed genes between samples.⁵Functional analysis is an essential component of gene

analysis, which aims to determine the roles and functions of genes. This can be done through laboratory experiments, such as gene knockout or gene overexpression studies in model organisms. Additionally, functional analysis can involve studying protein-protein interactions, protein function prediction, and pathway analysis to gain insights into the molecular mechanisms underlying biological processes.

Gene analysis has numerous applications across various fields. In medical research, it helps identify genetic risk factors for diseases, develop personalized treatments, and understand the molecular basis of diseases. Pharmacogenomics, a branch of gene analysis, studies how genetic variations influence an individual's response to drugs, aiding in personalized medicine. Gene analysis is also crucial in agriculture and crop improvement. It enables the identification of genes responsible for desirable traits in plants and animals, facilitating selective breeding and genetic engineering for improved productivity, disease resistance, and nutritional value. Furthermore, gene analysis contributes to forensic science by providing DNA profiling techniques for identifying individuals and establishing relationships between individuals in forensic investigations. In summary, gene analysis is a multidisciplinary field that involves studying an organism's DNA to gain insights into its genetic makeup, traits, and disease susceptibility.⁶ Through various techniques, such as DNA sequencing, genotyping, gene expression analysis, and functional analysis, gene analysis helps unravel the complex world of genetics, leading to advancements in medicine, agriculture, and forensic science. The increasing adoption of genomic medicine, advancements in DNA sequencing technologies, and the growing need for personalized medicine are some of the key factors driving the growth of the gene analysis market. Additionally, the decreasing costs of DNA sequencing and the availability of high-throughput sequencing platforms have made gene analysis more accessible to researchers and healthcare professionals. It's worth mentioning that the gene analysis market is highly competitive, with several established

companies and research institutions actively participating in this field. Key players in the market include Illumina, Thermo Fisher Scientific, Qiagen, Agilent Technologies, and BGI Group, among others.⁷As the field of gene analysis continues to evolve, with advancements in AI, machine learning, and big data analytics, it is expected that the market size will continue to grow. The increasing focus on precision medicine, the rise of direct-to-

consumer genetic testing, and the integration of genomics into routine healthcare are likely to contribute to the expansion of the gene analysis market in the coming years. According to data collected from various researches conducted by human genome project and Broad Institute of MIT and Harvard, the projection of gene analysis market size is given below:

YEAR	TOTAL MARKET SIZE OF GENE ANALYSIS (BILLION USD)
2021	9.80
2022	10.64
2023	11.56
2024	12.55
2025	13.63
2026	14.80
2027	16.07
2028	17.45
2029	18.95
2030	20.57

Table 1.

Given below is a graph displaying the above data which shows the market size will almost double itself in the year 2030:

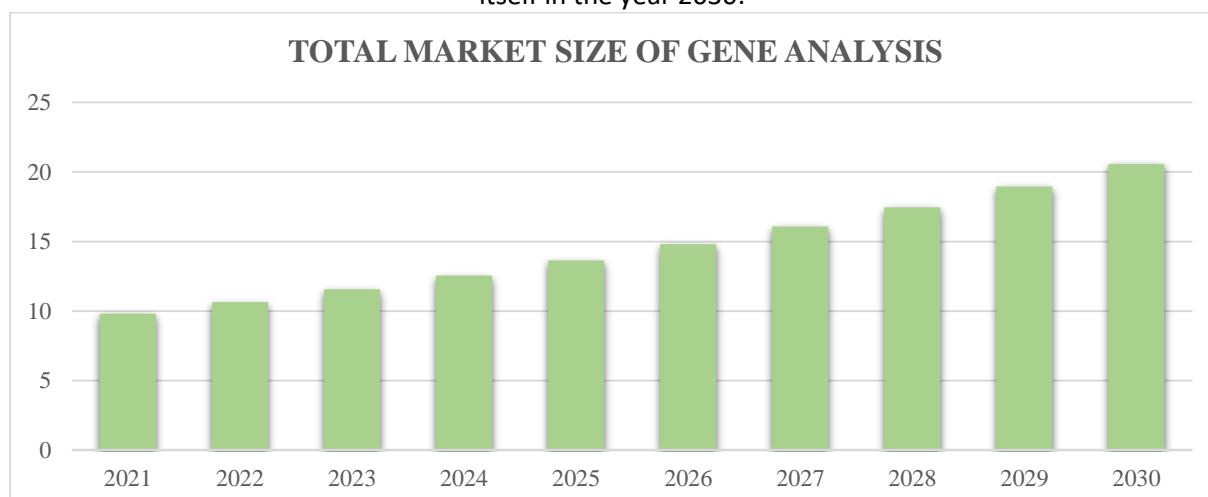


Fig. 1

ROLE OF ARTIFICIAL INTELLIGENCE IN GENE ANALYSIS:

Artificial Intelligence (AI) plays a crucial role in gene analysis by providing powerful computational tools and algorithms to handle

the vast amount of genetic data generated through DNA sequencing, genotyping, and gene expression analysis. AI techniques enable researchers to extract meaningful insights, discover patterns, and make



predictions about genetic information more efficiently and accurately. Here are some specific ways AI contributes to gene analysis:

1. Data analysis and interpretation: AI algorithms can process and analyze large-scale genetic datasets quickly, allowing researchers to identify patterns, genetic variations, and relationships that might be difficult to detect manually. Machine learning techniques, such as clustering, classification, and regression, can be applied to categorize and interpret genetic data, facilitating the discovery of genetic markers, disease associations, and potential drug targets.⁸

2. Genomic sequencing and assembly: AI algorithms can aid in the process of DNA sequencing and genome assembly, which involve piecing together short DNA fragments to reconstruct the complete genome sequence. AI-based algorithms can help optimize the sequencing process, improve accuracy, and reduce costs, making genomic sequencing more accessible and efficient.

3. Variant calling and annotation: Identifying genetic variations, such as single nucleotide polymorphisms (SNPs) or structural variations, is a fundamental task in gene analysis. AI techniques can be employed to develop algorithms that accurately detect and annotate genetic variants from sequencing data, distinguishing between benign and pathogenic variants and predicting their functional impact.

4. Predictive modeling and risk assessment: AI algorithms can be used to develop predictive models that estimate an individual's risk of developing certain diseases based on their genetic profile. By analyzing genetic data alongside other relevant factors, such as environmental exposures and lifestyle factors, AI can assist in personalized risk assessment,

enabling early interventions and tailored preventive strategies.

5. Drug discovery and development: AI has the potential to revolutionize drug discovery and development processes. By analyzing genetic data, AI algorithms can identify potential drug targets, predict drug efficacy, and assist in designing novel therapeutic interventions. AI can also facilitate virtual screening of large compound libraries to identify molecules with potential therapeutic effects, accelerating the drug discovery process.

6. Precision medicine: AI enables the integration of genetic data with other clinical and demographic information to support personalized medicine approaches. By analyzing a patient's genetic profile, AI algorithms can aid in predicting treatment responses, selecting appropriate therapies, and minimizing adverse effects, leading to more effective and personalized medical interventions.

7. Image analysis: In gene analysis, images such as microscopy images, histopathological slides, or imaging modalities like MRI or CT scans may be used to study genetic expression and cellular structures. AI algorithms can analyze these images to identify patterns, classify cell types, and detect abnormalities, enhancing the understanding of gene expression and cellular mechanisms.⁹

GENE ANALYSIS USING AI SYSTEMS: UNDERSTANDING THE PROCESS

AI systems used for gene analysis typically consist of several key components and architectural elements. The specific architecture may vary depending on the application and the complexity of the analysis being performed.

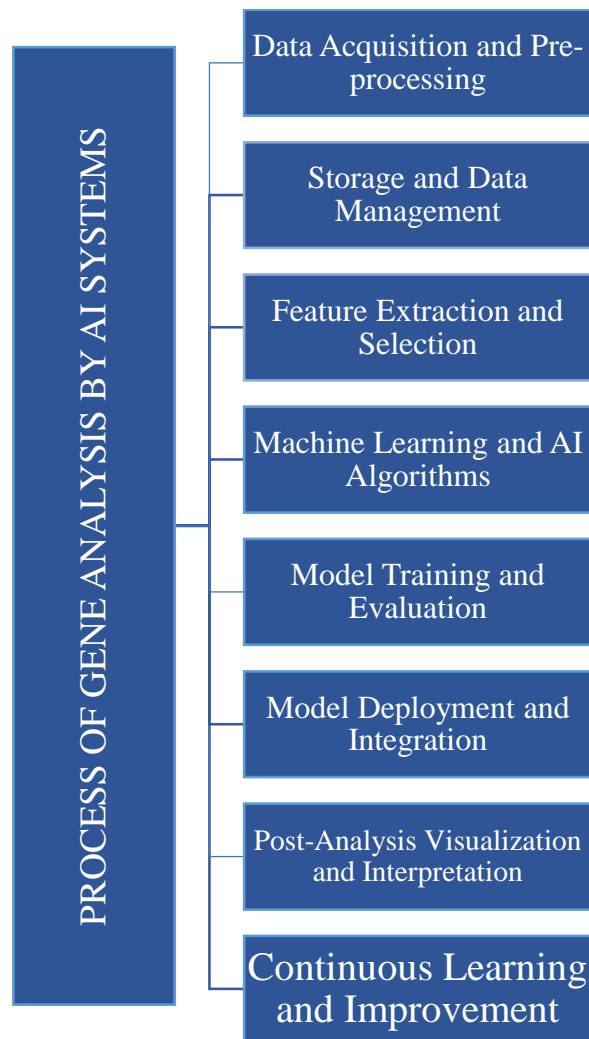


Fig. 2 Process of gene analysis using AI systems

The first step in gene analysis is the acquisition of genetic data, which can include DNA sequencing data, genotyping data, gene expression data, or other relevant datasets. This data is typically obtained from research studies, public databases, or clinical samples. Before analysis, the data undergoes pre-processing steps such as quality control, filtering, normalization, and data integration to ensure its accuracy and compatibility.³Genetic data is typically stored in databases or data repositories designed to handle large-scale genomic datasets. These systems provide efficient storage, retrieval, and management of genetic data, ensuring data integrity, security, and accessibility. In gene analysis, relevant features or attributes are extracted from the genetic data. This step involves identifying informative genetic markers, gene expression profiles, or other

features that are relevant to the analysis objectives. Feature selection techniques may be employed to reduce the dimensionality of the data and remove irrelevant or redundant features. AI algorithms, particularly machine learning algorithms, are applied to the pre-processed genetic data to extract patterns, make predictions, and derive meaningful insights. Various machine learning techniques, such as supervised learning, unsupervised learning, and deep learning, can be utilized depending on the specific analysis tasks. These algorithms learn from the data and make predictions or classifications based on learned patterns and statistical models. The AI system undergoes a training phase where the machine learning models are trained on a labeled dataset, typically using a subset of the available genetic data. During training, the models learn the relationships and patterns in

the data. The performance of the models is evaluated using appropriate metrics and validation techniques to assess their accuracy, precision, recall, or other relevant measures. Once the AI models are trained and evaluated, they can be deployed and integrated into the gene analysis pipeline. This may involve developing APIs (Application Programming Interfaces) or software interfaces to allow seamless interaction with other tools, databases, or platforms. The integration may also involve creating user-friendly interfaces for researchers or healthcare professionals to interact with the AI system. After the analysis, the results generated by the AI system are visualized and interpreted to gain insights and facilitate decision-making.⁴ Visualization techniques such as plots, charts, or interactive dashboards are employed to present the findings in a meaningful and easily understandable manner. AI systems used for gene analysis are often designed to continuously learn and improve over time. This may involve incorporating new data, updating models, or adapting to evolving genetic research and clinical knowledge. Feedback loops and monitoring mechanisms can be implemented to track the system's performance and make necessary adjustments or updates.

ADVANTAGES OF USING AI SYSTEMS FOR GENE ANALYSIS:

The use of AI systems in gene analysis offers several advantages that contribute to advancements in research, diagnosis, and personalized medicine. AI systems excel at processing and analyzing large volumes of genetic data quickly and efficiently. With the exponential growth of genomic data, AI algorithms can handle complex analysis tasks, such as variant identification, gene expression analysis, and pathway analysis, in a timely manner. This enables researchers and clinicians to explore vast datasets and extract valuable insights that may not be feasible with traditional analysis methods. AI algorithms can enhance the accuracy and precision of gene analysis. They can identify subtle patterns, correlations, and relationships within genetic data that may not be apparent to human

analysts. By leveraging machine learning and statistical techniques, AI systems can provide more accurate predictions, classification, and stratification of genetic variants, improving the understanding of disease mechanisms and guiding personalized treatment decisions.

AI systems can expedite the diagnosis process by automating the interpretation of genetic data and identifying disease-causing mutations or biomarkers. They can help clinicians in identifying genetic disorders, predicting disease progression, and selecting appropriate treatment strategies. AI systems also support the development of targeted therapies and personalized medicine approaches by matching patients with specific genetic profiles to the most effective treatments. AI systems can uncover novel genetic associations and identify potential disease risk factors that may have been overlooked by traditional analysis methods. By integrating diverse data sources, such as genomic data, clinical data, and population health data, AI algorithms can identify hidden patterns and complex interactions, contributing to the discovery of new biomarkers, drug targets, and therapeutic interventions.

AI systems enable predictive modeling and risk assessment by analyzing genetic data alongside other patient-specific information, such as medical history, lifestyle factors, and environmental influences. This enables the development of personalized risk profiles, early disease detection, and proactive interventions to prevent or mitigate the onset of diseases.⁵ AI systems can also support genetic counseling by providing accurate risk assessment and tailored recommendations based on an individual's genetic makeup. AI systems facilitate the integration of multi-omics data, including genomics, transcriptomics, epigenomics, and proteomics. By combining and analyzing data from multiple molecular levels, AI algorithms can provide a comprehensive understanding of the biological processes underlying diseases and identify potential therapeutic targets. This integrative approach enhances the depth and breadth of gene analysis, enabling a more

holistic view of complex genetic traits and diseases.

AI systems have the ability to continuously learn and improve over time. By leveraging machine learning techniques, AI algorithms can adapt to new data, refine their models, and enhance their predictive capabilities. This enables AI systems to keep pace with emerging research findings and evolving knowledge in the field of genetics, ensuring that gene analysis methods remain up-to-date and provide the most accurate and relevant insights. The advantages of using AI systems in gene analysis empower researchers, clinicians, and healthcare providers to gain deeper insights into genetic variations, accelerate diagnosis and treatment decisions, and drive advancements in precision medicine. AI systems complement traditional analysis methods by leveraging computational power, pattern recognition, and predictive modeling, revolutionizing the field of gene analysis and its impact on healthcare.

USE OF AI SYSTEMS FOR GENE ANALYSIS: THE FLIP SIDE OF THE STORY

While AI systems bring numerous benefits to gene analysis, there are also certain drawbacks that need to be considered. AI algorithms, particularly complex deep learning models, can be challenging to interpret. They often operate as black boxes, making it difficult to understand the underlying decision-making process. This lack of interpretability can hinder the ability to explain the reasoning behind the results generated by AI systems in gene analysis, which may be critical for clinical decision-making or research validation.

AI systems heavily rely on the quality and representativeness of the training data. If the training data is incomplete, biased, or contains errors, it can adversely impact the performance and accuracy of AI algorithms. Biases in the training data, such as underrepresentation of certain populations or genomic variations, can result in biased predictions or misleading conclusions. Careful data curation and mitigation of bias are crucial to ensure the reliability and fairness of AI-based gene analysis. The use of AI systems in

gene analysis raises ethical and privacy concerns. Genetic data is highly sensitive and contains personal and familial information. Adequate measures must be in place to protect the privacy and confidentiality of individuals' genetic information. Additionally, ethical considerations should address issues such as informed consent, data ownership, potential discrimination, and the responsible use of AI-generated insights. While AI systems can assist in gene analysis, they should not replace human expertise. Human interpretation, domain knowledge, and clinical judgment are still essential components in understanding and validating the results generated by AI systems. It is important to strike a balance between the capabilities of AI and the involvement of experts to ensure accurate and reliable interpretation of genetic data.

AI systems trained on specific datasets or populations may have limited generalizability to diverse populations or datasets with different characteristics. Models trained on one population may not perform as effectively when applied to other populations with distinct genetic variations or demographic profiles. Ensuring the robustness and transferability of AI models across different populations and datasets remains a challenge in gene analysis. Validating the performance and reliability of AI systems in gene analysis is crucial but can be challenging. The dynamic nature of genetic research and the continuous evolution of AI algorithms require rigorous validation processes to ensure the accuracy and clinical utility of AI-based findings. Additionally, complying with regulatory frameworks, such as data protection regulations and guidelines for clinical decision support systems, poses additional challenges in the deployment and adoption of AI systems in gene analysis.

AI systems in gene analysis may produce large volumes of data and results, which require careful interpretation and integration into clinical or research contexts. Proper training and support for users in understanding and effectively utilizing AI-generated insights are essential to derive meaningful conclusions and avoid misinterpretation or misuse of the

results. Addressing these drawbacks requires a multidisciplinary approach, involving collaboration between researchers, clinicians, data scientists, ethicists, and policymakers. By addressing the challenges associated with AI systems in gene analysis, it is possible to maximize the benefits while mitigating the risks and limitations, ultimately enhancing the application of AI in improving genetic understanding, diagnosis, and personalized healthcare.

The future prospects of gene analysis using AI systems are promising and hold significant potential for advancements in personalized medicine, drug discovery, and our understanding of genetic diseases. AI systems will play a vital role in advancing precision medicine approaches. By integrating genetic data with other clinical, environmental, and lifestyle factors, AI algorithms can enable the development of personalized treatment plans tailored to an individual's genetic makeup. AI can help identify specific biomarkers, predict treatment response, and optimize therapeutic interventions for improved patient outcomes. AI systems have the potential to revolutionize the process of drug discovery and development. By leveraging large-scale genomic and molecular data, AI algorithms can identify novel drug targets, predict drug efficacy, and optimize drug design. AI-driven approaches, such as virtual screening and de novo drug design, can accelerate the identification and optimization of potential drug candidates, leading to more efficient and targeted drug development pipelines.⁶ AI systems will continue to facilitate the integration and analysis of multi-omics data, including genomics, transcriptomics, epigenomics, proteomics, and metabolomics. By combining and analyzing data from multiple molecular levels, AI algorithms can uncover complex interactions, identify disease mechanisms, and provide a comprehensive understanding of genetic traits and diseases. This integrative approach will contribute to advancements in personalized medicine and the identification of novel therapeutic targets. AI systems will further enable seamless integration and sharing of genetic data across different research and clinical settings.

Advanced AI techniques, such as federated learning and secure data sharing frameworks, will allow researchers and clinicians to collaborate and leverage diverse datasets while ensuring privacy and data protection. This will promote knowledge exchange, increase the statistical power of analyses, and facilitate large-scale genomic studies. There is a growing focus on developing explainable AI models and improving the interpretability of AI-generated results in gene analysis. Future prospects include the development of AI systems that provide transparent explanations of their decision-making processes, enabling researchers, clinicians, and patients to understand and trust the results. Explainable AI will be essential for integrating AI systems into clinical practice and fostering wider adoption. AI systems will increasingly serve as real-time clinical decision support tools in gene analysis. By continuously analyzing and interpreting genetic data, AI algorithms can provide clinicians with actionable insights, treatment recommendations, and risk predictions at the point of care. This real-time support can aid in faster and more accurate diagnoses, treatment decisions, and monitoring of genetic conditions. AI systems in gene analysis will be seamlessly integrated with other digital health technologies, such as wearable devices, mobile apps, and electronic health records. This integration will enable continuous monitoring of genetic data, patient-reported outcomes, and environmental factors, allowing for more comprehensive and personalized health assessments. AI-driven algorithms can analyze these integrated datasets to identify patterns, predict disease risks, and provide personalized recommendations for disease prevention and management. AI systems will continue to improve through continuous learning and adaptation. By leveraging techniques such as reinforcement learning and active learning, AI algorithms can continuously update their models, refine predictions, and incorporate new scientific findings. This iterative learning process will ensure that AI systems remain up-to-date and provide accurate and relevant insights as the field of genetics evolves.

Overall, the future prospects of gene analysis using AI systems are exciting, with the potential to revolutionize healthcare, accelerate scientific discoveries, and improve patient outcomes. Continued research, technological advancements, and collaborations between AI experts, geneticists, clinicians, and policymakers will be crucial in realizing the full potential of AI in gene analysis.

CONCLUSION:

In conclusion, gene analysis using AI systems represents a significant leap forward in our understanding of genetics, personalized medicine, and the development of novel therapeutics. The integration of AI algorithms and techniques in gene analysis has brought numerous advantages, including the ability to process large volumes of genetic data, improve accuracy and precision, expedite diagnosis and treatment, and uncover novel genetic associations. AI systems enable researchers and clinicians to explore complex genetic variations and identify crucial biomarkers that contribute to disease onset, progression, and response to treatment. Despite the numerous benefits, there are also challenges and limitations associated with the use of AI systems in gene analysis. These include the limited interpretability of AI models, concerns about data quality and bias, ethical and privacy considerations, the need for human expertise, and the validation and regulatory compliance of AI-driven findings. Addressing these challenges requires a multidisciplinary approach, involving collaboration between experts in genetics, AI, ethics, and regulation. Looking ahead, the future prospects of gene analysis using AI systems are highly promising. Advancements in precision medicine, drug discovery, and the integration of multi-omics data are anticipated.

AI systems will continue to play a pivotal role in personalized medicine, enabling the development of tailored treatment plans based on an individual's genetic profile. Moreover, the integration of AI with other digital health technologies and the continuous learning and adaptation of AI algorithms will

enhance real-time clinical decision support and provide more comprehensive and personalized patient care.

To fully realize the potential of gene analysis using AI systems, ongoing research and development are necessary. Further improvements in interpretability, transparency, and fairness of AI models are needed to build trust and facilitate widespread adoption. Additionally, collaborations and data-sharing initiatives will promote the integration of diverse datasets, ensuring representativeness and robustness in AI-driven gene analysis. In conclusion, gene analysis using AI systems has revolutionized the field of genetics, offering unprecedented opportunities for personalized medicine, advancing our understanding of genetic diseases, and accelerating therapeutic discoveries. By harnessing the power of AI, we can unlock the full potential of genetic data and pave the way for a future where healthcare is increasingly tailored to the individual, leading to improved patient outcomes and a deeper understanding of the intricate complexities of the human genome.

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