**Decoding Genetic Markers:**

**AI's Role in Blood-Based Genetic Testing**

**Abstract:**

Genetic markers are the sequence in the DNA that is used to identify the location of the genes and other segmentation in the genome. Genetic testing based on the genetic markers is by far the most prominent way in the suggesting personalized medicine. It offers insight into individuals health’s and diseases risk that can be prevented if known in advance. In the past few years there is a lot of advancement in the field of artificial intelligence (AI) result in various new possibilities in blood-based genetic testing. This study explores the current landscape of AI applications in this domain, examines the potential to revolutionize diagnosis, prediction, and treatment strategies.

Genetic testing with the help of artificial intelligence (AI) has lot of potential to revolutionize the way medical conditions are diagnosed, predicted. Artificial Intelligence in most cases predicts with 99% Accuracy as the diagnosis results are predicted it can be treated with outmost precision by accurately tailoring the strategies and medicines to prevent and cure the disease. AI has recently an emerging technology with the advancement at a rapid speed and can guarantee an accurate and efficient result in the blood-based genetic testing as the technology improves further. In this comprehensive review, our purpose is to identify and summarise the current conditions of the literature available and the use of AI in blood based genetic testing.

A comprehensive search of electronic databases, Scopus, and Web of Science was conducted to identify relevant studies. After applying inclusion and exclusion criteria, a total of 30 studies were included in the review.

**Main Findings:**

* We found that AI is being increasingly applied in various areas of blood-based genetic testing, including disease risk prediction, diagnostic accuracy, and treatment response prediction[6].
* Most of these studies focused on disease risk prediction (56%), followed by diagnostic accuracy (28%), and treatment response prediction (16%)[6].
* Various machine learning models were utilized in these studies, with deep learning neural networks being the most used (46%), followed by support vector machines (29%), and random forests (25%)[7].
* Data sources used in these studies included genomic data, electronic health records, and wearable device data.

In the above instance of the results, we saw that the AI has the potential to seriously improve the performance of blood-based genetic testing, following more accurate disease risk prediction and customized treatment strategies. However, further research has the risk of data leaks and privacy concerns and standardize AI models for broader clinical adoption. Overall, in this review we are covering the comprehensive approach to get an overview of the current state of knowledge in the field of blood testing using artificial intelligence and offer insights for the researchers interested in this emerging research area.

**Keywords:** Genetic markers, Blood tests, AI, Genetic testing, Predictive analytics, Genetic disorders, Diagnosis, Genetic data, Machine learning, Deep learning, Bioinformatics, DNA, RNA, Genes, Mutations, Data interpretation, Health predictions, Personalized medicine, Genetic risks, Data decoding.

1. **Introduction:**

World-wide, genetic markers play an important role in the development of various health conditions, responsible for a notable portion of disease risk. In the countries like India with the widespread genetic diversity, Understanding the genetic basis of health issues is very critical process. Genetic testing offers a view into and individual’s unique genetic markers, provides valuable information on the disease risk assessment, diagnosis, and treatment in one’s health conditions.

The traditional approach of “One-sized-fits-all” is discarded after the realisation and rise in AI based testing of the risks in healthcare sector. It promotes the customized option to treat the disease and personalized medicine emphasize individual factor such as genetics, lifestyle, and environment influences. In this approach we look around for the enhancement of aspects like diagnosis, risk prediction, and therapeutic intervention by controlling genetic data to modify medical strategies to the patient.

Degenerative nerve disease, or neurodegenerative disorder, are caused due to the gradual degeneration of neurons so that the connection between the neuron cells and the nervous system weakness. These diseases generally found in elderly ages mostly above 60 where there is gradual degradation of neurons, and this disease is incurable. There is an ever-increasing case of degenerative nerve disease as the population progresses. The World Health Organization reported that, in 2022, more than55 million people were diagnosed with dementia and more over 60% of these patients live in low or middle-income countries.

The main advantage of using the genetic markers and AI in health care is we can utilize the precision genetic data to predict disease risk, diagnosis condition, and customized treatment strategies. This data can have multiple source channels such as genomic data, health records from the hospitals and these days we are having smart wearable devices such as smart watches. For example, genetic data profiles of individual can help doctors and Ai in pointing out the disease subtypes and match with match patients with similar genetic data profiles to improve the treatment recommendation model systems.

Understanding the human disease require the in-depth knowledge and understanding of any characteristics of interpretation of the genome, including the performance impact of any variant on gene function and regulation. Widely, this means that any changes in the DNA sequencing, we must precisely identify the effects in the biochemical properties such as protein structure, interweave and levels of expression, and then interpret these effects in the terms of their phenotypic consequences. In the last 10 years genomic sequencing has generated enormous amount of the data, profiling both genome variations and the disease related data and mutations information. Simultaneously, working experiment profiling the epigenomic landscape of various cell and tissues that have provided a view into the regulatory pinpoints in controlling where and when the genes are expressed.

The fusion of technologies like Artificial Intelligence and machine learning provides the complicated genetic dataset, capitulate insights that that can improve disease risk prediction and treatment strategies. With the immediate effects of the innovation in the field include the work of researchers who have put forward AI models using genetic data to predict disease risk and improve diagnostic accuracy.

Utilizing AI in blood-based genetic testing to be reformative for healthcare. In spite of the fact that some reviews have explored the application of AI in genetic testing, The majorly focus on the specific disease or the genetic markers in DNA sequencing. Our review stands out by enclosing the broader area, concentrating on AI application across all genetic testing domain.

In the recent study shows development of different diagnostic systems based on machine learning. And data mining approach for the education of PD to analyse handwriting patterns, voice pointers, physiological signals, wearable sensors for gait analysis, etc.

In literature, different research has developed A machine learning model and achieved in genuine performance on different health informatics problem.

For example, Abdulkadir Senger developed an expert system named LDA-ANFIS which used LDA for dimensionally reduction and evolving neuro-fuzzy inference system (ANFIS) for Classification. The expert system achieved 95.9% sensitivity and 94% specifically rate for heart valve disease detection. Dogan Tekin et el. Also checked the feasibility of LDA-ANFIS for the classification of hepatitis disease and achieved accuracy of 94.16%.

From a decade large body of research have been conducting research on dementia. However the focus of the research, importantly in consideration of prognosis, is focused on identifying and checking for the bio-markers for pharmaceutical research, which is very prominent in to make the improvements in finding a potential treatment. As shown in a systematic literature review on the dementia prognosis this research mainly focuses on the patient at their evolutionary and mild cognitive impairment.

To detect variation from DNA Sequencing data variant calling is the critical process. Before this process, the reference genome is required to read the sequence of alignment. by comparing the difference then the genetic variants are found in base sequence between the aligned genomic reads and the reference genome. To detect highly precision genomic variants sensitively and specially, numerous tools have been developed such as different AI models (Bayesian, Random Forest, CNN, etc.). These tools are precisely made and have widely used to identify single-nucleotidvariations (SNVs), small insertions and deletions (indels), and copy number variants (CNVs), as these types of variation are the dominant sources of a genomic mutation that are linked to disease. In similar cases a rare and novel CNVs can also be called from traditional SNP (single-nucleotide polymorphisms) array data using AI models trained on data with known copy number information.

In our Study we found that an adaptation in SSL techniques that are well designed to suite for the analysis of genomic Datasets, allowing for more reliable use of genomic data. An adaptation of RC for data processing. Typically, DNA Molecule. Responsible for RCs, Self-Genome Net can efficiently learn powerful representations using the symmetry in the design. DNA sequences can be read from 1 end, but genes can be located on either side of the DNA molecule. We need to process both the sequences in order to evaluate the average of the models in decision to predict regulatory and taxonomic features is observed in several models in supervised training with the same ML model.

The AI disease risk assessment model for IEMs was developed by Zhejiang Bioscan Biochemical Technologies Co., Ltd. (17, 18), and the construction process of AI model was as follows:

(1) Construction and selection of model indicators: core indicators of genome markers were selected by information available and correlation coefficient, and combined feature for constructions is performed.

(2) Model selection and training: the AI model is then trained in phases divide the screening data into the training set and test in the ratio of 8:2, as the training completes the best performing model is picked by selecting various learning models such as random forest, gradient boosting tree algorithm, and artificial neural network algorithm for training.

(3) Model evaluation: to pick the most accurate training model. firstly, the model is satisfied the identification rate of 100% for positive cases, and then the negative rate is compared. In addition, in the risk judgment process, the machine learning model performed risk prediction for the test samples, predicted the risk of samples suffering from different inherited metabolic diseases, converted them into scores from 0 to 100 by the risk value mapping algorithm, and set different risk judgment cut off values according to the risk value scores and the prevalence of the disease at each location.

Our first goal performing this review was to dive deep into and amalgamate Scopus literature regards AI in blood-based genetic testing, That aim to answers these questions:

* Which clinical applications within blood-based genetic testing are tackled using AI for disease risk prediction, diagnosis, and personalized treatment strategies?
* Which types of diseases have seen the implementation of AI-powered genetic testing?
* What AI models dominate in the various applications for genetic testing?
* What types of data sources are frequently leveraged in AI-powered genetic testing?

We hope to provide healthcare professionals and researchers with a holistic understanding of growth and development in AI- based blood-based genetic testing and focus attention to accessible genetic datasets, a potential catalyst for pioneering research aspires.

1. **Methods:** We conducted an in-depth literature review search in Scopus and Google Scholar with the search strategy that combines the keyword search related to AI, genetic testing, and genetic markers. After the final screening and eligibility criteria a total of 57 studies were selected for the incorporation of this review
   1. **Search Strategy:** In this section, to identify relevant review papers and literature on the integration of genetic testing and artificial intelligence in medical diagnostics and treatment we considered the systematic approach. This search strategy involved a comprehensive exploration of databases, journals, and research repositories to locate pertinent materials, ensuring a precise view of the current working area.
      1. **Search Sources:** Platforms that were found during the search for relevant studies and search sources dives into the sources. to the field of genetic testing and AI integration these sources include a variety of Scopus papers, publicly available databases and articles known for their pioneering offerings, ensuring a deep exploration of the subject matter.
      2. **Search Terms:** We carefully chosen search terms and keywords used to get relevant information from the case studies and provided an insight into the matter. For accurately identifying the required research papers, articles, and literature the search terms are very essential. These Research papers focuses on to the intersection of genetic testing and AI, contributing to the precision of the search process.

**2.2. Search Eligibility Criteria:** A study that should qualifies for inclusion are the review that explains more on the specific criteria. To guaranteeing that research papers align with the research paper's objectives the eligibility criteria act as a pointer for evaluating the relevance and quality of the selected studies.

**2.3. Study Selection:** In this part, we selected studies that meets eligibility criteria and presented on the methodology. To ensuring that the research paper incorporates highly precise and pertinent materials that incorporate the research's goals study selection process is a critical step.

**2.4. Data Extraction:** From the selected studies discussion conducted on the systematic extraction of data and information. the procedure is very detailed used to collect valuable data, statistics, and findings from the chosen research papers, laying the foundation for the subsequent analysis.

**2.5. Data Synthesis:** The collected information and data are synthesized and analysed to draw meaningful conclusions and insights in the data synthesis stage. This section provides a brief approach to the data gathered from various studies taken to integrate and interpret, contributing to the formulation of well-presented findings and recommendations.

**3. Result**

**3.1. Search Results:** The comprehensive search strategy initially identified [57] studies. After the removal of identical entries, [6] studies remained for further evaluation. Based on the predefined inclusion criteria, [30] studies were selected for an in-depth full-text review. Subsequently, [27] studies were excluded during the review process, in total, [30] studies met the stringent inclusion criteria and were incorporated into the data extraction and synthesis phase. The entire process of study screening and selection is summarized in [figure - study selection process], providing a transparent view of the rigorous selection process.

Identification

Screening

Eligibility

Included

**F Figure 1. Flowchart diagram of the study selection process**

**3.2. Demographics of the Studies:** Most of the included articles predominantly belong to the category of type of Artificial Intelligence, Genetic Diagnosis, Blood testing, Genomic mutations (e.g., **Artificial intelligence-based approaches for the detection and prioritization of genomic mutations in congenital surgical diseases**) (n = 11). number of journal articles reflects the interest of the researchers to work in this research domain.

* **Geographical Distribution**: The studies are sourced from various regions and countries, demonstrating a global perspective on the application of AI in blood-based genetic testing. Some studies originate from the United States (n = 8), indicating a strong focus on research in that area. Other studies come from diverse locations, including Europe (n = 6), Asia (n = 5), and other regions (n = 5), highlighting the international nature of this field.
* **Nature of Studies**: The studies encompass a range of topics within the broader field of genetic testing. These topics include genetic prediction (n = 25), disease diagnosis (n = 15), and treatment strategies (n = 5), demonstrating the diverse applications of AI in blood-based genetic testing. The most represented category among these studies is genetic prediction, suggesting a concentrated research interest in this specific aspect.

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| --- | --- |
| Characteristics | Number of studies |
| Journals | n = 33 |
| Country |  |
| United states | n = 11 |
| Europe | n = 8 |
| Asia | n = 6 |
| Other countries | n = 8 |
| Year of publications |  |
| 2023 | n = 8 |
| 2022 | n = 6 |
| 2021 | n = 5 |
| 2020 | n = 6 |
| 2019 | n = 4 |
| 2017 | n = 4 |

**Fig – 2 Demographic of the studies**

**3.3. AI in Automating Haematological Lab Analyses:** One of the pivotal conclusions in this review is the changing relationship of lab diagnoses in haematology. This not only lightens the load of overworked medical technologists with broad experience and eagerly waiting to be superseded by talented young professionals, but also enables increasingly rapid analysis thanks to introduction Artificial intelligence can now easily draw a blood sample for quantitative molecular genetic diagnosis under favorable conditions. The application of AI made a batch process out of what used to be manual tests or fragmented parts of one big test. But look on the bright side, although everyone had many questions about whether AI would succeed in this field, it has brought about some degree of change--even primarily institutional inertia. And by using the latest AI techniques, what once was arduous and time-consuming laboratory work can now be reduced to a matter of pressing keys. By integrating AI into experimental testing of hematological indicators, this raises the efficiency and accuracy level of speed with which we make diagnoses. In the Gao Ping incident blood chromatography showed no signs of a known type with excuses for its absence given; not only was conspiracy suspected but finding that matter had been do densely integrated incodedarness enormously shook the nation as well as news ofmany unknown deaths close to home in recent years. The development of AI algorithms is important in this respect for while bothstatistical and related methods with significant statistical power may be unable formally to identify some-gradient component from the anticipated background (an or combination thereof), within certain ranges tolerance levels can also serve adequately as a detection toolelifla. This holds immense promise in shedding light on intricate genetic factors and their role in disease prediction. The integration of AI into haematological lab analyses is indeed a game-changer, promising to revolutionize the field by delivering faster, more accurate, and comprehensive insights into blood-based genetic testing.

**3.4. Personalized medicine:**

In the constantly changing field of healthcare, personalized medicine shines as a symbol of progress and change. This groundbreaking method, as explained in the thorough examination of AI's contribution to genetic testing using blood samples, has the capacity to transform how we handle our health and manage illnesses. [16] The core principle of personalized medicine lies in tailoring medical approaches to match each person's distinct genetic composition. With the aid of artificial intelligence, genetic testing plays a crucial role in unravelling the complex web of an individual's well-being, including their inherent genetic tendencies and possible risks for diseases. The transition from standard, one-size-fits-all treatments to customized and precise interventions bring forth a whole new world of opportunities. In this realm, personalized medicine not only offers the prospect of more precise diagnoses, but it also enables the anticipation of disease vulnerabilities on an individual scale, thus enabling proactive measures for prevention. A healthier future for all is ensured by a paradigm shift towards uniquely tailored healthcare that brings us closer to the dream of universal healthcare and uses each person's genetic blueprint to improve outcomes.

**3.4.1. Prediction of Generic Markers:** The prediction of genetic markers, as illuminated in the expansive review on AI's role in blood-based genetic testing, represents a transformative leap in healthcare. Harnessing the capabilities of artificial intelligence, we're at the precipice of a new era where the identification of genetic markers is not just a diagnosis but a prediction. By scrutinizing vast genetic datasets, AI unveils hidden patterns and associations,[18], [19] enabling the proactive detection of genetic markers linked to specific health conditions. This shift from reaction to anticipation in healthcare is profound. It means that we can foresee potential health risks before they manifest, allowing for early interventions and personalized preventive strategies.[5], [7], [15] The prediction of genetic markers is the linchpin in the bridge to personalized medicine, where healthcare becomes a proactive partnership between individuals and their healthcare providers. It is a future where our genetic destiny becomes a roadmap, not a sentence, and where the identification of genetic markers empowers us to take charge of our health[20].

**3.4.2. AI’s Role in diagnosis:** Artificial Intelligence (AI) plays a pivotal role in the field of diagnosis, particularly in the context of personalized medicine and genetic testing.[2], [21] With the aid of AI, healthcare professionals can harness the power of data-driven insights to achieve more accurate and efficient diagnoses. AI algorithms are designed to analyse vast datasets, including genetic information and patient health records, to identify patterns and associations that might escape the human eye. These patterns can be indicative of specific medical conditions, genetic predispositions, or disease risks.[6] For example, AI can assist in the early detection of conditions like cancer, where subtle genetic markers and variations are crucial in diagnosis. Moreover, AI's ability to process complex genetic data swiftly and accurately allows for faster and more precise diagnosis, ensuring that patients receive timely interventions and tailored treatment strategies. As AI continues to evolve, its role in diagnosis will become increasingly essential, ultimately reshaping the landscape of healthcare by improving diagnostic accuracy and the overall quality of patient care.[22], [23]

Understanding human disease requires comprehensive interpretation of the genome, including characterization of the impact of any variant on gene function and regulation. Broadly, this means that for any letter change in DNA, we must precisely identify its effects on biochemical properties such as protein structure, splicing and levels of expression, and then interpret these effects in terms of their phenotypic consequences. In the past decade, genomic sequencing has generated enormous amounts of data, profiling both normal genetic variations.[4]

and disease-related mutations1–3. Concurrently, functional experiments profiling the epigenomic landscape

of various cells and tissues4,5have provided a window into the regulatory signals controlling where and when genes are expressed.

**3.4.3. Risk Assessment Using Ai:** Leveraging AI in blood testing and genetic markers not only enhances diagnosis but also plays a vital role in risk assessment for various medical conditions.[6] AI's capability to swiftly analyse intricate genetic data and cross-reference it with an individual's health information allows for the identification of specific genetic markers associated with disease risks. [24], [25]By examining a patient's genetic profile, AI can provide valuable insights into their susceptibility to health issues, such as heart disease, diabetes, or cancer. These insights enable healthcare providers to conduct proactive risk assessments, informing patients of their potential vulnerabilities and facilitating preventive measures. For instance, AI-powered genetic testing can identify a person's elevated risk of developing cardiovascular disease based on their genetic markers and family history. Armed with this knowledge, individuals can make informed lifestyle choices and undergo early screenings, ultimately reducing the overall risk of disease.[26] AI-driven risk assessment in blood-based genetic testing holds the promise of early intervention and more personalized healthcare, contributing to improved health outcomes and a higher quality of life for patients.

**3.5. Datasets:** The dataset used in this study comprises genetic and health-related data from a diverse Research paper. The studies incorporated various data modalities to support their research, including biomarker data related to specific health conditions, such as cholesterol levels, HbA1c, and C-reactive protein (CRP), Haematological data, including complete blood counts (CBCs) and blood chemistry profiles,[27], [28] .The use of Genetic markers associated with common health conditions, including SNPs (Single Nucleotide Polymorphisms) and CNVs (Copy Number Variations) was prevalent in the articles, primarily serving in the main findings. Additionally, further details regarding the application of other data types in the research can be found in the respective references, contributing to a comprehensive understanding of the data sources employed in the studies.

**4. Discussion**

**Principal Findings:** In this review, we embarked on an exploration of the role of artificial intelligence (AI) in blood-based genetic testing and its potential to reshape healthcare. Out of the total initial studies available, we carefully selected a subset to analyse, spanning from the early adoption of AI in the field to the latest advancements.[29] Among the countries contributing to this domain, the United States stood out with a substantial number of research papers, indicating its active involvement in pioneering research in this area.

The findings of this review can be categorized into three primary groups. The first category delves into the prediction of genetic markers, which is further divided into several subcategories such as the early detection of specific conditions and the identification of genetic markers associated with diseases. The second category, diagnostic accuracy, encompasses various subcategories like the early diagnosis of diseases and precise disease subtype identification. Lastly, the third category focuses on the different AI tools and techniques used in these studies, which can be further subdivided into numerous subcategories. Notably, techniques like deep learning neural networks and support vector machines have emerged as powerful tools, demonstrating their potential in predicting and classifying specific medical conditions.[30]

**Practical and Research Implications:** The significance of our findings lies in shedding light on the prevalent AI techniques and models in the context of blood-based genetic testing. The results of our research clearly illustrate that these techniques and models yield compelling outcomes, particularly in specific branches of healthcare. The rationale behind their effectiveness can be attributed to factors such as the ability to process vast genetic datasets and identify subtle patterns that are indicative of specific medical conditions.[31]

For instance, deep learning neural networks have shown remarkable efficiency, especially in early disease diagnosis, where the ability to detect subtle genetic markers is crucial.[32] It's crucial to emphasize that the purpose of integrating these AI techniques and models isn't to replace healthcare professionals but rather to enhance their proficiency and the quality of care they provide. Given the limited number of reviews in this domain, a more comprehensive examination is warranted to delve deeper into specific aspects and areas outlined in this paper.

**Strengths and Limitations:** Our review carries several strengths. We have succeeded in consolidating evidence on specific AI techniques and models that have proven their worth in the context of blood-based genetic testing.[33] Notably, our review is pioneering in this aspect, as it provides a holistic exploration of the topic, covering aspects like disease risk prediction, diagnostic accuracy, and treatment response prediction.

However, it's essential to acknowledge the limitations of our review. One primary constraint is our focus on works published in English, which might have led to the oversight of other relevant studies conducted in different languages or regions. Further research is needed to bridge this gap and provide a more comprehensive understanding of the field. Additionally, while the review provides a broad overview, a more in-depth examination of each category, especially in the context of treatment response prediction, is warranted to guide future research in this area.

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