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RESEARCH PAPER

Leveraging Generative AI for Precision Medicine: Interpreting Immune Biomarker Data from EHRs in Autoimmune and Infectious Diseases

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ABSTRACT

This study reviews the interpretation of immune biomarkers by generative artificial intelligence regarding precision medicine. Precision medicine customizes diagnostics and therapy based on various individual characteristics of patients, especially genetic and immune biomarkers in autoimmune and infectious diseases. Generative AI manages these in much-simplified ways through real-time decision-making in the improved clinical outcome by analyzing a complex Electronic Health Records system. During PRISMA guidelines, the systematic review published 655 articles filtered to study 40 articles pertaining to generative AI in immune biomarker analysis and electronic health records. These studies used machine learning (ML), generative adversarial networks (GANs), transformers, and large language models (LLMs). Generative AI makes a stride ahead in real-time biomarker analysis to predict risk, efficacy in treatment, and vaccine design for diseases such as lupus, rheumatoid arthritis, sepsis, and COVID-19. The challenges are data inconsistency, ethical matters, and AI interpretability. It is of paramount importance to improve data standardization, systematized AI as application, interdisciplinary collaboration, and great enhancement in way of effective application in generative AI with precision medicine.

Keywords: Generative AI, Precision Medicine, Immune Biomarkers, Electronic Health Records (Ehrs), Autoimmune Diseases, Systemic Review

Introduction

Precision medicine brings revolution in healthcare, focusing on solely tailoring treatment based on individual characteristics, such as genetic profiles, environmental factors, and lifestyle choices. Originally discussed in autoimmune and infectious diseases, the difficulty of patient responses is an added challenge (Lam et al., 2021). Immune biomarkers-bodily particles akin to cytokine, autoantibody, and genotype markers-constitute a crucial part in both diagnosis and managing strategies, and their scattered effectiveness for treatment only goes further to compromise established treatment modalities (Restrepo et al., 2016).

Electronic Health Record (EHR) data consists of a wealth of health data spanning longitudinal capture from laboratory results and historical treatments; however, EHR data are voluminous and intricate and necessitate high-degree analytics tools for transformation into usable insights. The generative AI, on the other hand, with its capacity to see complex patterns and predict possible scenarios, is quite a grand solution in healthcare analytics, paving the way for actual real-time decisions and personalized intervention (named Ahmed, 2022).

Integrating Generative AI into Precision Medicine could turn out to be considerably invaluable toward better patient outcomes. With immune biomarker data obtained from electronic health records, Generative AI can then extrapolate the disease progression, predict the response to treatment, and identify novel therapeutic modalities (Shah et al., 2023). Such applications are essential for autoimmune and infectious diseases given the power to timely and accurately interpret the biomarker data, which has to be essential for correct treatment.

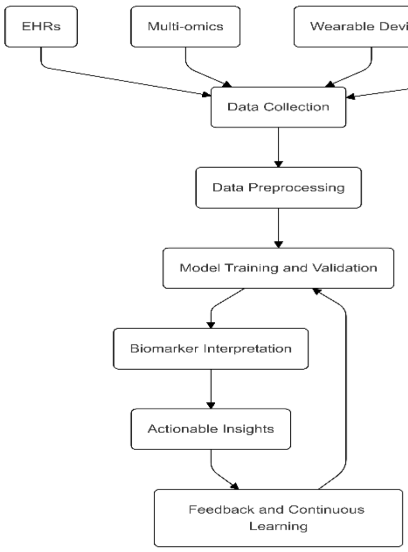

Figure1: Workflow of Generative AI in Immune Biomarker Analysis

Synergistic benefits notwithstanding, a variety of practical difficulties make it hard to decode immune biomarker data in real-time. The high dimensionality and heterogeneity biomarkers often face when analyzed by classical methods are associated with unsatisfactory results and delayed decision-making (Zucco, 2022). Moreover, interoperability and the quality of various data types like genotypic, proteomic EHRs and clinical team require some advanced computational techniques for integration into multi-modal data (Chin & Rider 2022). Generative AI a viable answer but there are many gateways to get over while incorporating it in clinical practice. Privacy and bias in AI models are a big challenge but also the trust of clinicians in decisions fomented with AI. Furthermore, the absence of suitable evaluations frameworks to automatically assess the performance of AI models interpretating immune biomarker data hinders their wider application (Yang et al., 2024).

Literature Review

Generative AI (GAI) is setting up a change in precision medicine; the tasks include analyzing various variables, including complex immune biomarkers and electronic health records (EHRs) for disease prediction and treatment personalization. Precision medicine attempts to customize mechanisms of treatment on the basis of patient-specific data, genetic information, and immune markers (Shah et al. 2023). However, huge amounts of unstructured EHR data put urgent demands on AI-based analytics for real-time insight (Ahmed 2022).

AI in Immune Biomarker Analysis

Biomarkers such as cytokines, autoantibodies, and genetic markers are necessary for good diagnosis and management of autoimmune and infectious diseases (Restrepo et al. 2016). Generative AI merges multi-omics data to increase biomarker discovery and predictive modeling (Shiwlani et al., 2024). Recent advances in ML, GANs, transformers, and

LLMs have improved biomarker interpretation, modeling disease progression, and making clinical decisions (Yuan et al. 2023).

Applications with respect to Autoimmune and Infectious Diseases

Uses AI in carrying out predictive models for premature disease onset and optimum biological therapies in autoimmune diseases such as rheumatoid arthritis and lupus (Basha & Hanirex, 2024). For similar reasons, AI applies to infectious diseases like COVID-19 and sepsis in areas of risk stratifying, treatment efficacy, and vaccine development (Bello et al., 2023). Development of AI biomarkers has enhanced immune response detection, inflammatory marker detection, and therapeutic outcome assessment (Rahman & Schellhorn, 2023).

AI-Related Challenges for Precision Medicine

Issues include inconsistencies, ethical challenges, and the interpretability of AI. Overall, the fragmentation of EHR data among different healthcare institutions impacts the accuracy of AI; hence, creating platforms and standards for data sharing and distribution is required (Sadeghi et al., 2024). Similarly, AI transparency remains a concern because many models function as black boxes, which retards the trust in the application. Ethics, with respect to patient privacy and algorithm bias, should call for solid AI governance and compliance with regulations (Zucco, 2022).

Generative AI, on the other hand, is expected to drive precision medicine, but may face challenges in clinical adoption: greater data integration, regulation, ethical considerations and transparency in AI models. Future research needs to focus on improving the reliability of AI, enhancing the interdisciplinary collaborative practice mechanism and developing regulatory guidelines to make it work effectively in healthcare.

Material and Methods

This systematic review was conducted following the PRISMA 2020 guidelines to evaluate the application of Generative AI for real-time interpretation of immune biomarker data from Electronic Health Records (EHRs) in the context of precision medicine. The review focused on autoimmune and infectious diseases. The timeframe for included studies was from 2015 to January 2024, and the process involved four key steps: identification, screening, eligibility, and inclusion.

Step 1: Identification

The identification phase utilizes comprehensive searches in major databases, including PubMed, Google Scholar, and IEEE Xplore, to ensure a wide scope of coverage. The search strategy employed Boolean operators to refine results, focusing on studies relevant to generative AI applications in immune biomarkers and precision medicine. The search string used was:

("Generative AI" OR "AI-driven interpretation" OR "machine learning") AND ("immune biomarkers" OR "biomarker data" OR "immune response") AND ("electronic health records" OR EHRs) AND ("precision medicine" OR "personalized medicine") AND ("autoimmune diseases" OR "infectious diseases").

This process yielded 655 articles, which were compiled for the subsequent steps in the review.

Step 2: Screening

The screening process involved an initial review of titles and abstracts by two independent researchers to assess relevance. Studies included if they explicitly addressed the use of generative AI or machine learning for interpreting immune biomarkers, integrating EHR data, or advancing precision medicine. Papers were excluded if they did not focus on generative AI or precision medicine, or if they addressed diseases outside the scope of autoimmune and infectious diseases.

In cases of disagreements between the reviewers, discussions were held to achieve consensus. When necessary, a third reviewer was consulted. This phase resulted in a subset of 500 papers deemed relevant for detailed eligibility evaluation.

Step 3: Eligibility Criteria

Eligibility criteria were established a priori to ensure the inclusion of high-quality, relevant studies. (Brony et al., 2024) These criteria are summarized in Table 1.

Table 1
Eligibility Criteria for Review

Criteria	Inclusion	Exclusion
Timeframe	Studies published between 2015 and January 2024	Studies published before 2015
Peer-Reviewed	Only peer-reviewed articles	Non-peer-reviewed articles, preprints
Focus Area	Generative AI applications in precision medicine	Studies unrelated to generative AI
Language	English or translatable into English	Non-translatable languages
Disease Focus	Autoimmune and infectious diseases	Other disease domains

Step 4: Inclusion

The final 40 articles underwent comprehensive data extraction and synthesis. Each article was reviewed to extract the research objectives, AI methodologies employed, Types of datasets utilized, Performance metrics (e.g., accuracy, sensitivity, specificity), and Clinical relevance and implications of findings.

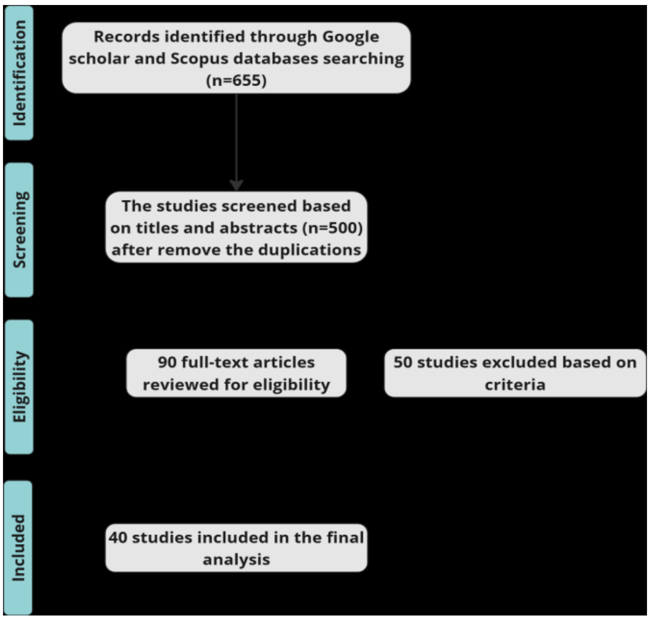

Figure 2: PRISMA Flowchart for Study Selection

Databases and Search Strategy

To ensure thoroughness, Boolean operators were employed to refine results and focus on studies relevant to autoimmune and infectious diseases. The search strategy aimed to capture every possible relevant study by including variations in terminology related to generative AI, immune biomarkers, and precision medicine. Details of the search process and the keywords used are summarized in Table 2.

Table 2

Summary of Search Strategy and Keywords

No.	Construct	Search Field/Limits
#1	"Generative AI" OR "AI-driven interpretation" OR "machine learning"	TS=Topic
#2	"immune biomarkers" OR "biomarker data" OR "immune response"	TS=Topic
#3	"electronic health records" OR EHRs	TS=Topic
#4	"precision medicine" OR "personalized medicine"	TS=Topic
#5	"autoimmune diseases" OR "infectious diseases"	TS=Topic
#6	2015–2024	PY=Year Published
#7	#1 AND #2 AND #3 AND #4 AND #5	Language: English

Search Methodology

The search process consisted of three stages: (Jiaqing et al., 2023; Brony et al., 2024)

Initial Search: A comprehensive database search was conducted to generate an initial pool of potentially relevant articles.

Screening: Titles and abstracts were screened to determine alignment with inclusion criteria.

Full-Text Review: Full-text articles were reviewed for methodological rigor, relevance, and completeness.

Data Extraction and Analysis

An analysis inspired by Dharejo et al. (2023) synthesized findings on the transformative role of generative AI in healthcare, particularly in interpreting immune biomarkers for precision medicine. Data extraction followed a standardized approach to ensure consistency, capturing key details such as study objectives, AI methodologies (e.g., machine learning, GANs, transformers, large language models), datasets, and performance metrics like sensitivity, specificity, accuracy, and area under the curve (AUC). Quantitative analysis highlighted trends, such as the superior predictive accuracy of generative AI models using GANs and transformers for multi-modal datasets (Sadeghi et al., 2024; Zucco, 2022).

Qualitative analysis identified common challenges, including biases in training data, limited model interpretability, and difficulties in clinical integration (Ahmed, 2022; Yuan et al., 2023). Findings were synthesized into actionable insights, addressing research gaps and emphasizing the importance of improving model explainability and scalability for broader adoption of AI in clinical settings, as highlighted by Terranova and Venkatakrishnan (2024). This review combines quantitative and qualitative analyses to provide a comprehensive overview of generative AI's potential in advancing precision medicine applications.

Results and Discussion

Table 3

Comparative Analysis of Key Studies on Generative AI in Precision Medicine

Author(s) and Year	Title	Study Focus	Methodology	Key Findings	Conclusion
Basha, S. A., &	Enhancing Immunological	Application of machine learning	Utilized ML techniques	ML models significantly	ML-based methods can

Hanirex, D. K. (2024)	Disorder Recognition through Machine Learning	(ML) for identifying and categorizing immunological disorders.	including deep learning, support vector machines, and random forests.	improved the accuracy of immunological disorder diagnosis and categorization.	revolutionize the detection and management of immunological disorders, offering precise and timely care.
Ajayi, A. F., et al. (2018)	Application of Data Science Approaches to Investigate Autoimmune Thyroid Disease in Precision Medicine	Data science applications in diagnosing and treating autoimmune thyroid diseases.	Analyzed big data approaches incorporating imaging, genetic sequences, and environmental risk factors.	Highlighted how genetic data and precision medicine tools enhance diagnosis and treatment of thyroid dysfunctions.	Precision medicine, when combined with AI and big data, significantly improves clinical autoimmune thyroid disease management.
Naik, K., et al. (2024)	Current status and future directions: The application of artificial intelligence/machine learning for precision medicine	Exploring AI/ML advancements and challenges in precision medicine, particularly in oncology and biomarkers.	Reviewed applications presented at a public workshop; discussed regulatory and technical challenges.	AI/ML innovations, like federated learning and synthetic data, improve biomarker discovery and patient care but require robust regulatory support.	Collaboration and regulatory frameworks are essential to maximize AI/ML's potential in precision medicine.
Restrepo et al., 2016	Shared genetic etiology of autoimmune diseases in patients from a biorepository linked to de-identified electronic health records	Shared genetic etiology of autoimmune diseases.	EHR-linked biorepositories were analyzed.	Identified shared genetic factors across multiple autoimmune diseases.	EHR-based genetic studies offer significant insights into autoimmune disease etiology, paving the way for more targeted genetic research and personalized treatment approaches.
Shiwlani et al., 2024	AI-Assisted Genotype Analysis of Hepatitis Viruses: A Systematic Review on Precision Therapy and Sequencing Innovations	AI applications in genotype analysis of hepatitis viruses.	Systematic review of precision therapy and sequencing innovations.	AI enhances genotype analysis and supports personalized therapy for hepatitis.	AI-assisted analysis provides significant advancements in hepatitis treatment and research.
George et al., 2024	Classification Framework for Autoimmune Liver Disease using Machine Learning and Deep Learning Techniques	Machine learning frameworks for autoimmune liver disease classification.	Proposed ML and DL-based classification framework.	Improved accuracy in diagnosing autoimmune liver diseases using ML techniques.	ML frameworks can support clinical diagnostics for autoimmune liver diseases.
McFadden et al., 2023	Developing machine learning systems worthy of trust for infection science: a requirement for future implementation into clinical practice	Trustworthy ML systems for infection science.	Review of ML system requirements for clinical practice adoption.	Trust and interpretability are critical for ML implementation in infection science.	Building trustworthy ML systems is essential for clinical integration.
Raikar et al., 2023	Advancements in artificial intelligence and machine learning in revolutionising biomarker discovery	AI advancements in biomarker discovery.	Explored ML techniques for biomarker analysis.	AI has revolutionized biomarker discovery for precision medicine.	AI-driven biomarker analysis enhances diagnostics and therapeutic strategies.
Krantz et al., 2022	Novel analysis methods to mine immune-mediated phenotypes and find genetic variation	EHR-based analysis of immune-mediated phenotypes and genetic variation.	Developed novel analysis methods for phenotype-genotype studies.	Identified genetic variations linked to immune-	EHR mining provides valuable insights into immunogenomics.

	within the electronic health record			mediated conditions.	
El-Sayed & Saber, 2022	Implementing Big Data-Driven Precision Medicine for Improved Clinical Outcomes in East Asia	Big data-driven precision medicine in East Asia.	Explored precision medicine applications using big data analytics.	Big data enhances clinical outcomes and precision medicine practices.	Adopting big data analytics accelerates advancements in precision medicine.
Macias et al., 2023	Utilizing big data from electronic health records in pediatric clinical care	Big data use in pediatric clinical care.	Analyzed EHR-based big data applications in pediatrics.	Big data improves decision-making and personalized care for pediatric patients.	EHR-driven big data analytics supports better outcomes in pediatric care.
Jeyaraj & Narayanan, 2023	Role of artificial intelligence in enhancing healthcare delivery	AI's impact on healthcare delivery.	Reviewed AI applications across healthcare systems.	AI enhances efficiency in diagnosis and treatment delivery.	AI is essential for modernizing healthcare services.
Yuan et al., 2023	Large language models illuminate a progressive pathway to artificial healthcare assistant: A review	LLMs as healthcare assistants.	Reviewed the use of LLMs in clinical contexts.	LLMs facilitate clinical decision-making with EHR analysis.	LLMs hold promise as transformative tools in healthcare, enabling complex data interpretation, improving diagnostics, and bridging gaps in clinical decision-making.
Cruz Navarro et al., 2022	A precision medicine agenda in traumatic brain injury	Precision medicine for traumatic brain injury.	Explored precision strategies for brain injuries.	Precision medicine aids in managing complex brain injuries.	Tailored strategies improve patient recovery outcomes.
Sarvan & Lakshmi Prasanthi, 2021	Personalized medicine: A new normal for therapeutic success	Personalized medicine in therapy success.	Reviewed PM strategies for therapeutic advancements.	Personalization improves therapeutic outcomes.	PM sets a new standard in healthcare effectiveness.
Khanna & Jones, 2023	Toward personalized medicine approaches for Parkinson disease using digital technologies	PM for Parkinson disease via digital tech.	Reviewed digital innovations in Parkinson care.	Digital tools enable tailored Parkinson treatments.	Tech-driven PM enhances Parkinson care.
Ho et al., 2020	Enabling technologies for personalized and precision medicine	Tech innovations enabling PM.	Analyzed advancements in PM-enabling tools.	Tech innovations drive PM advancements.	Adoption of new tools accelerates PM benefits.
Rahman & Schellhorn, 2023	Metabolomics of infectious diseases in the era of personalized medicine	Metabolomics in infectious disease PM.	Explored metabolomics applications in PM.	Metabolomics enhances infectious disease management.	Metabolomics is key in PM advancements, providing essential insights for understanding infectious diseases and enabling targeted interventions.
Bello et al., 2023	Integrating AI/ML models for patient stratification leveraging omics dataset and clinical biomarkers from COVID-19 patients: A promising approach to personalized medicine	AI/ML in COVID-19 patient stratification.	Developed AI/ML models integrating omics data.	AI enhances patient stratification in COVID-19.	AI-driven stratification improves COVID-19 care.

Begoli et al., 2018	Precision Medicine as an Accelerator for Next Generation Cognitive Supercomputing	PM in cognitive supercomputing.	Explored PM's role in AI advancement.	PM accelerates supercomputing innovations.	PM is pivotal for next-gen AI tools.
Li et al., 2023	Medical image analysis using deep learning algorithms	Deep learning for medical imaging.	Reviewed DL applications in imaging.	DL improves medical image diagnostics.	DL algorithms advance imaging-based diagnostics by improving the accuracy and efficiency of medical image analysis, leading to better patient outcomes.
Bao, Y. et al. (2024)	The UBA1–STUB1 Axis Mediates Cancer Immune Escape and Resistance to Checkpoint Blockade	Investigating the role of the UBA1–STUB1 axis in cancer immune escape mechanisms and its impact on resistance to checkpoint blockade therapies.	Experimental and computational analysis of the UBA1–STUB1 axis to understand its role in immune escape and resistance mechanisms.	The UBA1–STUB1 axis plays a critical role in immune escape, contributing to resistance against checkpoint blockade therapies.	Understanding the UBA1–STUB1 axis offers new avenues for developing therapies that overcome immune escape and resistance to immunotherapies. This axis could serve as a critical target for enhancing the efficacy of cancer immunotherapies, addressing the current limitations of checkpoint blockade treatments.
Choi, J. E. et al. (2024)	PIKfyve, expressed by CD11c-positive cells, controls tumor immunity	Exploring the regulatory role of PIKfyve in tumor immunity, specifically within CD11c-positive immune cells, to uncover potential immunotherapeutic targets.	Experimental study on CD11c-positive cells to analyze PIKfyve's role in controlling tumor immunity using immunological assays.	PIKfyve regulates tumor immunity and represents a potential target for enhancing anti-tumor immune responses.	Targeting PIKfyve in CD11c-positive cells can improve immune responses against tumors, opening new possibilities for cancer immunotherapy. This discovery highlights the potential of PIKfyve as a therapeutic target to enhance anti-tumor immunity and develop more effective cancer treatments.
Thatoi, P. et al. (2023)	Natural Language Processing (NLP) in the Extraction of Clinical Information from Electronic Health Records (EHRs) for Cancer Prognosis	Using Natural Language Processing (NLP) to extract clinical data from Electronic Health Records (EHRs) to improve cancer prognosis and enable more accurate decision-making.	NLP-based computational analysis of EHR datasets, focusing on data extraction and predictive modeling for cancer outcomes.	NLP techniques are highly effective in extracting and structuring EHR data for improved cancer prognosis and clinical decision support.	NLP can transform how clinical information from EHRs is utilized, leading to better cancer prognosis and treatment strategies. The ability of NLP to structure and analyze large-scale clinical data offers significant advantages in personalized cancer care and prognosis modeling.

Husnain, A. et al. (2024)	A Precision Health Initiative for Chronic Conditions: Design and Cohort Study Utilizing Wearable Technology, Machine Learning, and Deep Learning	Developing a precision health model that integrates wearable technologies with machine learning (ML) and deep learning (DL) to monitor and manage chronic conditions effectively.	A cohort study design leveraging wearable data and advanced ML/DL techniques to analyze chronic condition trends and predict outcomes.	The proposed model demonstrates high potential for real-time monitoring, trend prediction, and effective management of chronic conditions using wearable data.	Wearable technologies combined with ML/DL techniques provide actionable insights for precision health, improving outcomes for chronic disease management. This approach allows for real-time tracking of patient conditions, enabling timely interventions and optimizing healthcare delivery for chronic patients.
Shiwlani, A. et al. (2024)	Leveraging AI in Hepatology: Systematic Insights into Early Detection Models for Hepatitis-Linked Liver Cancer	This review investigates the application of artificial intelligence (AI) techniques for the early diagnosis of liver cancer caused by hepatitis, highlighting advancements in the field of hepatology.	A detailed analysis of AI models, including machine learning (ML) and deep learning (DL), focusing on their use in identifying early signs of hepatitis-associated liver cancer.	AI techniques showcased high effectiveness in detecting early stages of liver cancer, particularly in Hepatitis B and C patients, offering substantial potential for clinical adoption.	AI techniques showcased high effectiveness in detecting early stages of liver cancer, particularly in Hepatitis B and C patients, offering substantial potential for clinical adoption.

Discussion

Generative AI and Its Applications in Healthcare

Generative artificial intelligence (AI) is a fraction of machine learning dedicated to the provision of new content and the generation of predictive models. It works on the patterns already existing. AI for generative purposes, unlike traditional classification or regression AI, attains synthesis, forecasting, and decision-making as its high points, which are just suited to solve healthcare problems. It ensures that the healthcare system can manage complex and heterogeneous datasets, fulfilling the exploitation they are making in data interpretation for personalized medical care (Yang et al., 2024; Ahmed, 2022).

Definition and Types of Generative AI

Many generative AI models have been invented, each with their own unique set of peculiarities. With the help of Generative Adversarial Networks (GANs), which consist of a generator and discriminator, medical images and biological data are created to resemble reality and mitigate data scarcity for better predictive modeling (Peng et al., 2021). Transformers with BERT and GPT-3/9 are vital entities breaking sequential entities from analysis topics like EHR and drug discovery (Rane et al., 2023). With massive language models like GPT-4, applications for medical language comprehension have significantly improved to create electronic medical records and predict outcomes (Yuan et al., 2023).

The applications of generative AI in healthcare are data synthesis for filling data gaps due to missing values and privacy concerns, which are a must. This renders AI quite apt at prediction, tracing significant associations in datasets to forecast how any disease will proceed and the effectiveness of treatments (DeGroat et al., 2024). Responsible for proactive decision support in real-time, generative AI extracts constructive insights from EHRs so that clinicians are able to act in a timely and accurate way (Zucco, 2022).

Current Applications in Healthcare

The AI generation has emerged as a disruptive technology in all aspects of healthcare, providing new solutions to long-standing challenges and driving advances in healthcare practices.

AI in EHR Data Analysis

Various longitudinal datasets are available in Electronic Health Records (EHRs), which are a valuable source of patient information. The intricacy of these subjects frequently mandates the use of sophisticated analytical instruments to uncover valuable information. The completeness of data is enhanced by generating artificial intelligence models that synthesize missing data, which allows for improved patient histories. Moreover, these models promote collaboration by standardizing data formats across various healthcare systems to facilitate easy and efficient data integration and utilization. The use of artificial intelligence in pinpointing markers for disease development and treatment effectiveness is opening the field of precision medicine (Shah et al, 2023; Chin & Rider, 2002)

Use of Generative AI in Genomics and Biomarker Discovery

Generative AI has also proved useful in genomics and the discovery of new markers.' This allows researchers to synthesize genomic data and identify variants associated with infectious diseases or autoimmune diseases. This aptitude speeds up the recognition of disease mechanisms and facilitates targeted therapies (Restrepo et al, 2016).) Also, generative AI allows for the analysis of larger datasets (proteomics and transcriptomic studies) in search for novel biomarkers that can be used as indicators or predict diagnostic/therapeutic actions. In drug discovery, AI can predict molecular interactions and evaluate drug efficacy, which has a significant impact on reducing development timelines and costs (Ahmed, 2022; Moingeon, 2003).

Immune Biomarker Data and EHRs

The use of biomarkers is thus important in diagnosing, prognosticating, and managing a wide range of autoimmune and infectious diseases. Biomarkers, which can be proteins, autoantibodies, or acute-phase reactants, reflect immune system activity, which helps to monitor disease progressions and therapeutic responses of the disease (Restrepo et al., 2016; Ahmed, 2022). In general, biomarkers such as IL-6 and CRP are highly usable in monitoring inflammation in both autoimmune and infectious diseases, while ANA is essential in diagnosing SLE (Stafford et al., 2020). In infectious diseases, immune biomarkers allow for early detection and personalized interventions that significantly improve patient outcomes. For example, during COVID-19, biomarkers such as D-dimer and ferritin levels were important in risk stratification and therapeutic decision-making (Lam et al., 2021). Advanced AI techniques have further enhanced biomarker acquisition and have advanced precision medicine. Electronic Health Records (EHRs) provide comprehensive data, including clinical notes, diagnostic reports, lab results, and treatment histories, supporting personalized medicine and autonomous disease management (Chin & Rider, 2022; Shah et al., 2023). However, analyzing EHR data is challenging due to variability, gaps, and inconsistent quality. Generative AI addresses these issues by integrating multi-modal data and using imputation techniques to handle missing data (Zucco, 2022). When combined with EHRs, immune biomarkers provide more accurate diagnoses and personalized treatment options, offering significant advancements in precision medicine (Yuan et al., 2023).

Table 4
Key Biomarkers Identified in Immune Diseases.

Biomarker	Associated Diseases	Role in Disease	Relevance to Precision Medicine
IL-1β (Interleukin-1 beta)	Rheumatoid arthritis, systemic inflammatory diseases	Pro-inflammatory cytokine contributes to inflammation and tissue damage.	Serves as a target for biologic therapies in autoimmune diseases.
VEGF (Vascular Endothelial Growth Factor)	Cancer, autoimmune diseases (e.g., lupus nephritis)	Regulates angiogenesis and vascular permeability; linked to inflammation and tissue remodeling.	Biomarker for disease activity and response to anti-VEGF therapies in autoimmune diseases.
IL-17	Psoriasis, ankylosing spondylitis, RA	Promote inflammation; plays a role in autoimmune and chronic inflammatory diseases.	Target for biologic therapies in autoimmune conditions like psoriasis and ankylosing spondylitis.
CD4/CD8 Ratio	HIV, autoimmune diseases	Reflects immune system status; imbalance associated with immunosuppression or overactivation.	Monitored for disease progression and therapy adjustments in immune-related conditions.

Challenges and Limitations

Data Challenges

One of the main issues in developing the NHCT ecosystem for precision medicine is the quality, completeness, and biases that are in EHR and biomarker data. The preservation of the integrity of the effectiveness of AI technology for predictions when inadequate or inaccurate inputs are involved in EHRs (Ahmed, 2022). In addition, biases in data collection, such as the underrepresentation of certain populations, can limit the generalizability of AI models.

Ethical and legal issues also complicate the integration of generative AI into healthcare. Patient privacy must be protected and compliance with data protection laws such as GDPR and HIPAA must be ensured. Finding a balance between data availability and privacy in AI education remains a major challenge (Zucco, 2022).

Technical Challenges

AI-first models are often plagued by problems of interpretation and trustworthiness. Clinicians cannot accept AI insights if they do not understand the implications of the decisions. It is important to improve the clarity of the model through the implementation of signal processing (Chin & Rider, 2022). Integration and integration into clinical workflows are major challenges. Implementing integrated AI solutions across multiple healthcare systems requires robustness and ease of collaboration, both of which are often lacking in today's environment (DeGroat et al. (2024).

Domain-Specific Challenges

The variability in immune responses across populations adds another layer of complexity. Generative AI models must account for genetic, environmental, and lifestyle factors that influence biomarker levels, necessitating diverse and representative training datasets (Sadeghi et al., 2024). Additionally, the generalization of AI models to varied clinical settings remains a challenge. Models trained on specific datasets may not perform optimally in different healthcare environments, underscoring the need for rigorous validation and adaptation (Peng et al., 2021).

Future Directions

Enhancing Data Infrastructure

Enabling data portals formerly is very crucial for generative AI that is the successful completion of the medical sector. Establishing interoperable electronic health record systems that are easy to share data will help to improve the quality and quantity of datasets (Ahmed, 2022). Thus, the inclusion of multi-omics data, for example, genomics, proteomics, and metabolomics, with EHRs will be a complete and detailed description of the patient's health and this will make it possible for professionals to prescribe accurate and personalized drugs (Yuan et al., 2023).

Advancing Generative AI Models

Interdisciplinary activities like AI research, health practitioners, and bioinformatics experts are very important in exploiting precision medicine innovation. Defining performance metrics and benchmarks for which evaluations provide groundwork for AI models' reliability will help them to be easily integrated in the clinical practice (Rane et al., 2023). Lastly, establishment of innovative and real-world projects, where academic and industry people are involved along with the healthcare providers will enable the development and use of creative and constructive AI products.

Collaboration and Standardization

It is the cooperation of AI researchers, clinicians, and bioinformaticians from different areas that are crucial in the pace of precision medicine. The regulation of benchmarks and assessment tools which will be consistent in the evaluation of the performance of AI models will facilitate the process of their translation into clinical practice (Rane et al., 2023). Besides that, the cooperation of academic institutions, healthcare organizations, and industry stakeholders will help in the development and spread of creative AI solutions.

Conclusion

Imbalance in generative AI unambiguously supports the future of individualized illness treatment through the interpretation of complex datasets, such as immune biomarker data extracted from Electronic Health Records (EHRs). As a new path towards good science, this review highlights the potential of AI for diagnosis, management, and treatment of autoimmune and infectious diseases. Generative AI is an enabler of timely detection and growth of personalized therapeutics by mashing data with a prediction of the outcome as well as support to real-time decision-making. In other words, with some additional evidence from generative AI forms such as cytokine profiles and some biomarkers in predicting liver diseases, such as rheumatoid arthritis, lupus, and inflammatory bowel disease have shown good results in terms of early and more accurate diagnosis. For certain infectious diseases such as tuberculosis, sepsis, and COVID-19, AI is critical for proper classification of these diseases, vaccine development, and real-time disease monitoring. Nevertheless, various constraints stand in the way: data quality, biasedness, patient privacy issues, as well as concerns related to scaling up of models and interpretability. Effective application of generative AI in the domain of rehabilitation, general medicine and patient navigation programs and in an ethics- and transparency-promoting manner becomes quite essential if its broader potential is to be safely realized. Strengthening data infrastructure by means of interoperable EHR systems and harmonization of multi-omic level data will play a crucial role. Transparency and bias mitigation are important for designing AI models to assist present in clinician trust and equitable healthcare delivery. The strengthening of the collaborative relationships among AI researchers, bioinformaticians, and clinicians is critical to surmounting these existing obstacles and thus fast-tracking adoption into the healthcare arena. Generative AI offers a unique opportunity to transform healthcare by leveraging immune biomarker data, paving the way for precise, personalized, and equitable medical care as a standard practice in the future.

Recommendations

The generative AI community should pay attention to the following key focus areas to Augment its utility in precision medicine. Firstly, improvement in the quality of data and standardization of electronic health record (EHR) data is important to derive trustworthy insight from an AI standpoint. This includes putting together a multi-omics approach (genomics, proteomics, and metabolomics) toward biomarker discovery and predictive modeling. Second, ethical issues surrounding patient privacy and algorithmic bias ought to be fine-tuned; therefore, commercial AI systems must have transparency in building and comply with all data protection regulations (HIPAA) and general rules (GDPR). Thirdly, the effortless interpretability and explainability of AI tools can build trust among clinicians and support their integration into real-world scenarios. This would include the development of user-centric AI applications but must be based on solid validation studies in multiple areas of clinical practice. Computational infrastructures that support the interdisciplinary interaction of AI scientists, bioinformaticians, and clinicians will help these groups develop rapidly and will build up an AI-led personalized strategy to treatment. Finally, a set of instructions needs to be given by lawmakers on the proper uses of AI in clinical settings so as to ensure safe, effective, and fair treatment of all patients.

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