

AI-Enabled Genomic Biomarkers: The Future of Pharmaceutical Industry and Personalized Medicine

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Abstract

The integration of artificial intelligence (AI) into genomics has ushered in a new era in pharmacy and personalized medicine. The current research delves into the transformative potential of AI-enabled genomic biomarkers and their profound implications for the future of healthcare. AI-driven advancements in genomics have revolutionized the way pharmaceuticals are developed and administered. By harnessing the power of machine learning algorithms, AI can analyze vast and intricate genomic datasets with unprecedented speed and precision, leading to the discovery of highly specific and individualized biomarkers. These biomarkers offer tailored insights into disease susceptibility, treatment response, and drug interactions, thereby facilitating a paradigm shift towards personalized medicine. The study highlights how AI-powered genomic biomarkers are poised to address the long-standing challenges of one-size-fits-all medicine. They enable healthcare practitioners to identify patient-specific genetic variations, enabling the customization of treatment regimens and pharmaceutical interventions. The result is not only increased treatment efficacy but also a reduction in adverse effects and healthcare costs. Furthermore, this paper explores the ethical and regulatory considerations surrounding AI-enabled genomic biomarkers, emphasizing the importance of data privacy, transparency, and informed consent. It discusses the need for interdisciplinary collaboration between pharmacists, geneticists, AI experts, and regulators to ensure the responsible integration of AI into pharmacy practice. AI-enabled genomic biomarkers represent a promising avenue for enhancing the precision, efficacy, and safety of pharmaceutical interventions. The research underscores the transformative potential of these biomarkers in reshaping pharmacy and personalized medicine, ushering in an era where treatments are as unique as the individuals they serve. The future of pharmacy is intertwined with the intelligent analysis of genomics, guided by AI, and its commitment to delivering truly personalized healthcare solutions.

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INTRODUCTION

Genomic Biomarkers are the Genetic Signposts of a person. Every person's body is like a complex recipe, and their DNA is the cookbook that holds all the instructions for how their body works. Imagine that within this cookbook, there are special markers, like post-it notes, highlighting important information. These markers are called genomic biomarkers, and they are a key part of understanding the person's health. Body's instruction manual is written in a special code, just like a secret language. This code, known as DNA, tells the body how to grow, work, and stay healthy. But sometimes, there can be little "typos" or changes in this code. These changes can be important clues for doctors and scientists to understand the person's health better. Genomic biomarkers are like little flags or signposts in DNA that signal something important pointing to certain traits or tendencies the person may have. They are specific spots in genetic code that can show if the person might be at risk for certain diseases, how the body responds to medicines, or even why the person might have a particular health condition. Genomic biomarkers are specific genetic characteristics or variations that can reveal important information about the health and how body responds to various factors.

Literature Survey

The current advancements in genomics and biotechnology hold great potential for the creation of biomarkers that can predict an individual's disease risk, facilitate early disease detection, enhance diagnostic categorization, and provide valuable information for personalized treatment. Biomarkers are biological indicators used to forecast disease risk, detect diseases at an early stage, guide treatment choices, and assess the effectiveness of therapeutic interventions. The Human Genome Project was strongly motivated by the goal of identifying and creating these biomarkers to support the realization of personalized, preventive, and predictive medicine[1]. The use of molecular biomarkers in disease diagnosis, monitoring, and drug treatment has grown significantly in recent years. Although only a limited number have gained FDA approval, many more are being studied in translational research and clinical trials. However, information about which biomarkers are used, their purposes, and relevance to specific conditions is often hidden in free-text descriptions on platforms like ClinicalTrials.gov, making it challenging for both humans and machines to access and analyze. To address this, a text mining strategy has been developed to identify and classify proteomic and genomic biomarkers in clinical trials. Over 3,000 biomarkers

associated with around 2,600 diseases have been identified, allowing for insights into their use across different therapeutic areas and their specificity. This data is now available through the Clinical Biomarker App, which aims to facilitate biomarker discovery, development, and application [2]. The COVID-19 pandemic has had devastating effects on both human lives and the global economy. While vaccinations have helped mitigate its spread, the unstable RNA sequences of SARS-CoV-2 continue to pose challenges, necessitating additional drugs. This study used transcriptomics RNA-Seq data from SARS-CoV-2-infected and control samples to identify 109 differentially expressed genes (DEGs) and subsequently pinpointed 10 key hub genes/proteins through protein-protein interaction (PPI) network analysis. Functional and pathway enrichment analyses of these hub-DEGs highlighted their significance in SARS-CoV-2 infections. Additionally, the study identified five transcription factors (TFs) proteins and six miRNAs as key regulators of these hub-DEGs. Docking analysis with SARS-CoV-2 3CL protease-guided 90 FDA-approved drugs revealed seven promising candidates, including Torin-2, Rapamycin, and Radotinib, which exhibited strong binding affinities against various receptor proteins associated with COVID-19. Molecular dynamics-based simulations further confirmed the stability of these drugs, suggesting their potential effectiveness against different SARS-CoV-2 variants [3].

Alzheimer's disease (AD) is a progressive neurodegenerative condition influenced by genetic variations and brain imaging findings. Brain imaging biomarker genomics is a recent technique that combines genetic data with multimodal brain imaging to uncover AD's underlying mechanisms and enable early detection. This review outlines the fundamental framework of brain imaging biomarker genomics and describes its two primary applications in AD research: (1) identifying new biomarkers and promoting mutual understanding and (2) utilizing machine learning methods in conjunction with brain imaging biomarker genomics for AD diagnosis and prognosis. The importance of this approach is emphasized, along with an examination of current methodologies' strengths and limitations, and suggestions for future research directions in this field [4].

Molecular profiling of lung cancer tumors plays a critical role in tailoring personalized treatment plans for each patient. This research provides an update on the significance of genomic testing in lung cancer, highlighting targeted treatment options and discussing emerging biomarkers and mechanisms of resistance. With a deep understanding of molecular biomarkers, testing protocols, and their impact on treatment decisions, nurses can effectively educate, and support patients and their families as clinical care increasingly incorporates molecular profiling into the

management of lung cancer[5]. Liver cancer is a significant global cause of cancer-related deaths, emphasizing the importance of early detection and treatment. Biomarkers hold promise in aiding the early diagnosis and management of liver cancer, but their effective identification and application pose challenges. Recent literature suggests that artificial intelligence (AI) is emerging as a valuable tool in this context. This review highlights the growing role of AI in liver cancer research, focusing on its potential for risk prediction, diagnosis, staging, prognostication, treatment response prediction, and recurrence monitoring through the facilitation of biomarker utilization [6]. Precision medicine has revolutionized healthcare by allowing early disease risk prediction and tailored treatment plans. It involves analyzing comprehensive patient data and various factors to distinguish between healthy and ill individuals, leading to better understanding of biological indicators of health changes. The combination of precision and genomic medicine with artificial intelligence (AI) holds immense potential for enhancing patient care. Genomic medicine is especially beneficial for patients with unique healthcare needs or atypical therapeutic responses. AI, through its computational abilities, aids in decision-making and learning, enhancing the role of physicians. High-throughput measurement of cell characteristics, such as gene regulation and protein-nucleic acid interactions, can serve as training data for predictive models, and the integration of large datasets and machine learning techniques is paving the way for more effective genomic medicine. This review article highlights the valuable contributions of machine learning algorithms in precision and genomic medicine [7]. Artificial intelligence (AI) involves the development of computer systems capable of performing tasks that typically require human intelligence. Recent advancements in AI, particularly in deep learning algorithms and the powerful GPUs that support their training, have sparked significant interest in medical AI applications. AI-driven computer vision methods are set to revolutionize image-based diagnostics in clinical settings, and other AI subtypes are showing promise in various diagnostic modalities. In clinical genomics, deep learning algorithms are employed to process intricate genomic datasets. This review outlines the types of problems AI systems excel at solving and details how clinical diagnostics can benefit from AI solutions. Additionally, it highlights emerging AI methods in clinical genomics, such as variant calling, genome annotation, variant classification, and phenotype-to-genotype correspondence. The review concludes with a discussion on the future potential of AI in personalized medicine, particularly for risk prediction in common complex diseases, along with the challenges, limitations, and biases that must be thoughtfully addressed for

the successful integration of AI in medical applications, particularly those involving human genetics and genomics data[8].

The biomedical field is experiencing a surge in data generation, thanks to advances in technology, while computational capabilities have grown in parallel for data analysis. These trends extend beyond genome sequencing to encompass all omics disciplines, emphasizing the need for multi-omics integration and AI-driven data interpretation. Specifically, in mass spectrometry-based proteomics, machine learning and deep learning are now capable of predicting experimental peptide measurements solely from amino acid sequences, enhancing analytical workflows' quality and reliability. Machine learning is also pivotal in biomarker discovery from proteomics data, often surpassing existing assays. However, considerations such as model transparency, explainability, and data privacy are critical when deploying MS-based biomarkers in clinical applications [9]. Recent advancements in prostate cancer research are revolutionizing diagnosis and treatment. Traditional methods, like Gleason scores and PSA levels, are being complemented by new molecular biomarkers identified through transcriptomics, genomics, and artificial intelligence (AI). These biomarkers, including mRNAs, microRNAs, and lncRNAs, promise earlier and more precise diagnoses, enhancing the prognosis and quality of life for patients. Bioinformatics tools, such as machine learning and deep learning, play a crucial role in processing and interpreting vast sequencing data. Moreover, cutting-edge techniques like Spatial Transcriptomics and Exome Sequencing are enabling earlier detection, while the synergy of AI with genomics is shaping personalized prostate cancer management [10]. Precision medicine has significantly improved health outcomes by enabling earlier diagnosis and more accurate prognoses, particularly for chronic diseases. It combines patient clinical data with multi-omics/genomic information to tailor treatments based on individual needs, diverging from the one-size-fits-all approach. The integration of artificial intelligence, healthcare practices, clinical genomics, and pharmacogenomics plays a pivotal role in advancing precision medicine. These fields collectively contribute to patient-specific care, shifting the focus from generalized symptom management to personalized healthcare solutions. While offering great promise in disease prevention and prediction, they also pose ongoing challenges that need to be addressed for their full potential to be realized[11]. Artificial intelligence (AI) has made significant strides in healthcare, driven by advances in software, hardware, and deep learning algorithms. AI is poised to revolutionize

clinical diagnostics, particularly in image-based diagnostics, and is showing promise in various diagnostic modalities. In clinical genomics, deep learning algorithms are used to process complex genomic data, offering potential in individualized medicine applications, including risk prediction for common complex diseases. However, the successful deployment of AI in medical settings, especially in human genetics and genomics, necessitates addressing challenges, limitations, and biases[12]. Artificial intelligence (AI) is increasingly applied to complex molecular datasets in genetics and genomics, challenging traditional statistical modelling approaches.

This special issue on "Artificial Intelligence in Genomics" addresses the need for AI algorithms to be interpretable, reproducible, automatic, and translatable. Among AI methods, deep learning techniques are gaining traction, particularly in scenarios with intricate patterns beyond the scope of standard modelling. Treppner et al.'s contribution focuses on deep generative models, offering insights into their use for extracting complex information from molecular data and emphasizing methods that enhance interpretability through relationships between latent variables and observed data [13]. This review explores the current applications of artificial intelligence (AI) in the realm of functional genomics, driven by the recent surge in AI capabilities, particularly in deep learning, and the availability of vast biological datasets. Functional genomics encompasses various disciplines that study DNA information, reversible DNA modifications, RNA transcripts, epitranscriptomics, proteomics, and metabolomics. These fields collectively generate a massive amount of biological data, surpassing even astronomy in the production of big data. The review not only outlines AI's role in functional genomics but also highlights associated ethical, legal, and economic considerations and underscores the importance of interpretability in AI-driven genomics research[14]. Polygenic cardiovascular diseases, caused by multiple genes working together, present diagnostic and management challenges. The field of cardiovascular medicine aims to understand how genetic variations contribute to the clinical diversity in these diseases. Recent advances in artificial intelligence (AI) and the availability of next-generation sequencing (NGS) offer new possibilities for complex genomic analysis. Integrating AI and genomics holds potential for a deeper understanding of polygenic cardiovascular diseases, better prognosis, and personalized medicine. This review explores current opportunities and limitations in genomics, introduces AI, and discusses the current applications, limitations, and future directions of AI in genomics research[15].

Types of Genomes and uses

Different types of genomic biomarkers exist and are useful in various ways, offering insights into an individual's health, susceptibility to diseases, and responses to treatments.

1. **Single Nucleotide Polymorphisms (SNPs):** SNPs are the most common type of genetic variation. They involve a single nucleotide change in the DNA sequence. SNPs can be associated with an increased or decreased risk of certain diseases, and they can influence an individual's response to drugs. Examples include the APOE gene variants associated with Alzheimer's disease risk and the CYP2D6 gene variants affecting drug metabolism. **Uses-** **Disease Risk Assessment:** SNPs can indicate an individual's predisposition to certain diseases. For example, specific SNPs are associated with an increased risk of conditions like heart disease, diabetes, and cancer. **Drug Response Prediction:** SNPs can help predict how a person will respond to medications, allowing for personalized treatment plans. For instance, certain SNPs influence how the body metabolizes drugs, affecting their efficacy and safety.

2. **Copy Number Variations (CNVs):** CNVs involve the duplication or deletion of relatively large segments of DNA. They can influence susceptibility to diseases like autism, schizophrenia, and certain cancers. **Uses-** **Disease Diagnosis:** CNVs can be responsible for genetic disorders like Down syndrome, and their detection aids in diagnosing such conditions. **Cancer Research:** CNVs in cancer cells can reveal important information about tumor progression and guide treatment decisions. Amplifications or deletions of specific genes can drive cancer growth and may be targeted with therapies.

3. **Microsatellites:** These are repetitive sequences of DNA, also known as short tandem repeats (STRs). Instabilities in microsatellites can be indicative of certain genetic disorders, such as Huntington's disease and certain types of cancer.

4. **Gene Expression Patterns:** Genomic biomarkers can include patterns of gene expression, which can provide insights into disease mechanisms and treatment responses. For example, specific gene expression profiles in breast cancer tumors can help predict the likelihood of recurrence and guide treatment choices. **Uses-** **Disease Understanding:** Gene expression biomarkers provide insights into how genes are active or silenced in specific diseases. This knowledge aids in understanding disease mechanisms and potential therapeutic targets. **Cancer Subtyping:** In cancer, gene expression patterns can help classify tumors into subtypes, which can

have different clinical implications and treatment responses.

5. **Mutation Analysis:** Certain mutations in genes are strongly associated with specific diseases. For instance, mutations in the BRCA1 and BRCA2 genes are linked to an increased risk of breast and ovarian cancer. **Uses- Genetic Disorders:** Mutations can cause genetic disorders like cystic fibrosis, Tay-Sachs disease, or sickle cell anaemia. Identifying specific mutations helps diagnose these conditions. **Cancer Identification:** Mutations in certain genes, such as the BRCA1 and BRCA2 genes in breast cancer, are associated with an increased cancer risk. Detecting these mutations allows for proactive measures like increased screening or preventive surgeries.

6. **Epigenetic Modifications:** Epigenetic changes, such as DNA methylation and histone modifications, can affect gene expression without altering the underlying DNA sequence. Aberrant epigenetic modifications are associated with various diseases, including cancer. **Uses- Disease Mechanisms:** Epigenetic biomarkers reveal how environmental factors influence gene activity. They offer insights into conditions like cancer, where epigenetic changes can play a critical role in tumor development. **Drug Development:** Understanding epigenetic modifications can lead to the development of drugs that target these changes, potentially reversing them and treating diseases.

7. **Tumor Mutational Burden (TMB):** TMB measures the number of mutations in a tumor's DNA. High TMB is associated with a better response to immunotherapy in some cancers.

8. **Pharmacogenomic Markers:** These markers predict an individual's response to specific drugs based on their genetic makeup. Examples include the HLA-B gene for predicting hypersensitivity reactions to certain drugs and the TPMT gene for determining the appropriate dosage of thiopurine drugs.

9. **Genetic Risk Scores:** These scores combine information from multiple genetic variants to assess an individual's overall risk for a particular disease. For example, polygenic risk scores are used to estimate a person's susceptibility to conditions like coronary artery disease.

10. **Disease-specific Mutations:** Some diseases have specific genetic mutations associated with them, such as the CFTR mutations in cystic fibrosis or the FMR1 gene mutations in fragile X syndrome.

11. **Liquid Biopsy Markers:** In cancer, genomic biomarkers detected in liquid biopsies (e.g., circulating tumour DNA) can provide real-time information about tumour mutations and treatment response.

These different types of genomic biomarkers collectively contribute to precision medicine and personalized healthcare -Risk Assessment: They help determine an individual's susceptibility to specific diseases, enabling preventive measures and early interventions. Treatment Selection: Genomic biomarkers guide healthcare providers in selecting the most effective treatments with fewer side effects based on a patient's genetic makeup. Disease Understanding: They enhance our understanding of disease mechanisms, aiding in research and drug development. Prognosis: Biomarkers can provide information about disease prognosis and predict the likelihood of disease recurrence. Monitoring: Biomarkers can be used to monitor treatment response and disease progression, allowing for timely adjustments in treatment plans. Overall, the diverse range of genomic biomarkers provides a comprehensive view of an individual's genetic profile and its relevance to health, disease, and personalized medical care.

Role of AI in genome biomarkers

Artificial Intelligence (AI) plays a significant role in advancing our understanding and utilization of genomic biomarkers in various ways. Genomic data is incredibly complex and vast. AI algorithms can efficiently manage and process this data, enabling researchers to analyze millions of data points quickly. Pattern Recognition: AI can identify subtle patterns and associations within genomic data that might be challenging for humans to detect. This can lead to the discovery of novel biomarkers or disease-related genetic variations. AI algorithms can assist in identifying genetic variants, including single nucleotide polymorphisms (SNPs), insertions, deletions, and structural variations, with high accuracy. Functional Annotation: AI can predict the functional consequences of genetic variants, helping researchers understand how specific genetic changes might impact health or disease. AI can calculate polygenic risk scores (PRS) by analyzing multiple genomic biomarkers simultaneously. These scores provide insights into an individual's genetic predisposition to specific diseases. AI-driven models integrate genomic, clinical, and environmental data to understand the complex interplay between genetic factors and diseases with multifactorial origins. AI analyzes a patient's genomic biomarkers to recommend the most suitable treatments, taking into account their genetic profile and the latest research findings. This can lead to more effective and tailored therapies. AI assists in drug discovery by identifying potential drug targets based on genomic data and predicting how specific drugs may interact with genomic biomarkers. AI enhances the efficiency of DNA sequencing processes, reducing costs and

turnaround times. It can help identify genetic variants accurately and at a faster pace. Variant Classification: AI helps in the classification of genetic variants, distinguishing between pathogenic variants and benign ones. This is particularly important for diagnosing genetic disorders. AI can provide real-time support to clinicians by analyzing patients' genomic data and offering treatment recommendations or risk assessments based on the latest scientific knowledge. Interpretation Assistance: AI tools can help healthcare providers interpret the significance of genomic biomarkers and their implications for patient care, ensuring more informed decision-making. AI integrates diverse data sources, such as genomic data, clinical records, imaging, and more. This comprehensive approach offers a holistic understanding of disease progression and treatment response. AI-driven analyses uncover novel insights by identifying previously unknown connections between genomic biomarkers and diseases. This leads to new avenues of research and potential therapeutic targets. AI serves as a powerful ally in genomics, accelerating our understanding of genomic biomarkers and their roles in health and disease. Its ability to process, analyze, and interpret complex genomic data contributes to precision medicine, personalized healthcare, and advancements in biomedical research.

AI algorithms that can be applied on genomic biomarkers

Several AI algorithms and techniques are suitable for analyzing genomic biomarkers due to their ability to process vast amounts of genetic data and identify meaningful patterns. Here are some AI approaches commonly used in genomics:

Machine Learning (ML) Algorithms like Random Forests: Random forests are used for classification tasks, such as distinguishing between disease and non-disease states based on genomic biomarkers. They are robust and can handle high-dimensional data. Support Vector Machines (SVMs) are useful for classification and regression tasks. They can be applied to identify relevant biomarkers associated with specific diseases or traits. Deep learning neural networks, including convolutional neural networks (CNNs) and recurrent neural networks (RNNs), can analyze genomic sequences, identify patterns, and predict outcomes. Dimensionality Reduction Techniques like Principal Component Analysis (PCA) reduces the dimensionality of genomic data while preserving important information. It is used for data visualization and feature selection. t-Distributed Stochastic Neighbor Embedding (t-SNE) is used for visualizing high-dimensional data, making it helpful for exploring the relationships between different genomic biomarkers.

Clustering Algorithms like K-Means can group similar samples or genomic biomarkers together, helping to identify subtypes of diseases or patterns in gene expression data. Hierarchical Clustering: This method builds a hierarchy of clusters, providing insights into the relationships between different genomic features or samples. Natural Language Processing (NLP) Techniques like Text Mining are employed to extract information from scientific literature and databases. They help researchers gather knowledge about specific genomic biomarkers, their functions, and associations with diseases. Reinforcement learning is used to design novel drug compounds by optimizing molecular structures based on genomic information. It can identify potential drug targets and compounds with desired properties. Hidden Markov Models (HMMs) are employed in sequence analysis to identify patterns or motifs within genomic sequences, such as promoter regions or binding sites. Sequence Alignment Algorithms like BLAST and Smith-Waterman are used to align genomic sequences, facilitating the comparison of genetic variations and mutations. Bayesian networks are used to model causal relationships between genomic biomarkers and diseases, helping researchers understand the mechanisms underlying genetic associations. Deep Neural Networks (DNNs) including deep autoencoders, can capture intricate relationships within genomic data, making them valuable for feature extraction and classification tasks. Cox Proportional Hazards Model, which is statistical model, often used in conjunction with AI, helps predict patient survival based on genomic biomarkers and clinical data. Ensemble Learning techniques like bagging, boosting, and stacking combine multiple AI models to improve predictive accuracy when analyzing genomic data.

The choice of AI algorithm depends on the specific analysis goals, the nature of the genomic data, and the type of genomic biomarkers under investigation. Researchers often employ a combination of these techniques to gain a comprehensive understanding of the genetic factors influencing health and disease.

Impacts of AI enabled genomic biomarkers

Pharmacology

AI analyzes patients' genomic data to identify specific biomarkers associated with drug response and adverse reactions. Pharmacists can use this information to customize medication regimens, dosages, and formulations, ensuring that treatments are tailored to individual genetic profiles for maximum efficacy and safety. Pharmacists can use AI-powered tools to select the

most appropriate medications for patients based on their genomic biomarkers. This reduces the risk of adverse drug reactions and minimizes the need for trial-and-error approaches, leading to more effective and efficient treatment strategies. AI helps pharmacists identify potential drug-drug interactions that may be influenced by a patient's genetic makeup. This knowledge allows pharmacists to make informed decisions when multiple medications are prescribed, minimizing the risk of harmful interactions.

Pharmacogenomic information provided by AI assists pharmacists in understanding how genetic factors influence drug metabolism and efficacy. This knowledge empowers pharmacists to make informed recommendations to healthcare providers and patients. Pharmacists can play a crucial role in patient education and counseling regarding the implications of genomic biomarkers on their medication. They can explain how genetic variations may affect treatment outcomes and advise patients on lifestyle modifications or alternative therapies if necessary. AI-powered systems can help pharmacists monitor patients' adherence to prescribed medications by providing insights into how genetic factors may impact treatment response. This information can be used to develop strategies to improve medication adherence. Pharmacists can use AI to identify eligible patients for clinical trials based on their genomic profiles. This streamlines the patient recruitment process and ensures that participants have a higher likelihood of responding positively to investigational drugs. AI can assist in tailoring drug formulations to match patients' genetic predispositions. This includes adjusting the composition, dosage form, or delivery method of medications to optimize therapeutic outcomes. Integrating AI into pharmacy workflow management systems can enhance efficiency and accuracy in medication dispensing, inventory management, and patient record keeping. This allows pharmacists to focus more on patient care and counseling. Pharmacists must ensure the secure handling and protection of patients' genomic data, addressing privacy concerns and adhering to regulatory requirements for data security and consent. The evolving role of AI in genomics requires ongoing education and training for pharmacists to stay up to date with the latest advancements and ethical considerations.

AI-enabled genomic biomarkers are reshaping the practice of pharmacy by making treatments more precise, safer, and patient-centred. Pharmacists are pivotal in leveraging these advancements to provide personalized care and ensure optimal medication outcomes for their patients.

Precision medicine

AI plays a pivotal role in advancing precision medicine through the integration of genomic biomarkers. AI analyzes an individual's genomic data to identify specific biomarkers associated with diseases or drug responses. This enables the development of highly personalized treatment plans tailored to a patient's genetic profile. It can predict a person's susceptibility to various diseases based on their genetic makeup. This allows for proactive disease prevention and early intervention strategies. AI accelerates drug discovery by identifying potential drug targets and predicting how patients with specific genomic profiles will respond to new medications. This reduces the time and cost of bringing new drugs to market. AI models can forecast how patients will respond to particular drugs or therapies, helping clinicians choose the most effective treatments while minimizing adverse reactions. It aids in the selection of participants for clinical trials by matching patients with specific genomic biomarkers to relevant trials, enhancing trial efficiency and success rates. By identifying genomic biomarkers associated with adverse drug reactions, AI helps prevent or mitigate harmful side effects, improving patient safety. AI-guided genomic biomarker analysis enables the development of targeted therapies that focus on the specific genetic abnormalities driving a patient's disease, enhancing treatment precision. AI integrates genomic data with clinical, environmental, and lifestyle data to provide a comprehensive view of a patient's health, facilitating more holistic and personalized healthcare decisions. It can continuously monitor genomic data to track disease progression and adjust treatment plans in real time, optimizing long-term patient outcomes. Precision medicine guided by AI can potentially reduce healthcare costs by avoiding ineffective treatments and hospitalizations, leading to better resource allocation. The ethical use of AI in precision medicine requires careful consideration of data privacy, consent, and the responsible handling of sensitive genetic information. The integration of AI into precision medicine necessitates ongoing education and training for healthcare professionals to effectively interpret and apply genomic insights. Developing regulatory frameworks that ensure the safety and efficacy of AI-driven precision medicine solutions is crucial to their widespread adoption. AI in conjunction with genomic biomarkers, empowers precision medicine by enabling individualized treatment, improving drug discovery, reducing adverse events, and enhancing overall healthcare outcomes. Its ability to analyze and interpret vast genomic datasets provides a foundation for a more personalized, effective, and efficient healthcare system.

Countries investing in AI based genome research

Countries around the world are benefiting from AI in genome biomarkers, and several of them are actively advancing research, healthcare, and biotechnology through the integration of AI and genomics. Some notable countries and regions include:

United States: The United States is a global leader in AI and genomics. Major institutions, such as the National Institutes of Health (NIH), are investing heavily in AI-driven genomics research. The country hosts numerous biotech companies and startups focused on personalized medicine and drug discovery. **China:** China has made significant strides in AI-powered genomics, with a focus on large-scale genome sequencing projects. The Beijing Genomics Institute (BGI) is one of the world's largest genomics centres, employing AI for various research applications, including disease diagnosis and precision medicine. **United Kingdom:** The UK is renowned for its genomic research initiatives, including the 100,000 Genomes Project. AI is integral to these efforts, aiding in the analysis of large genomic datasets and supporting personalized healthcare initiatives. **Canada:** Canadian research institutions and biotech companies are leveraging AI in genomics to enhance cancer research, disease diagnosis, and drug discovery. The country is fostering collaborations between AI experts and geneticists to drive innovation in healthcare. **Singapore:** Singapore has emerged as a hub for AI and genomics research. The country's investments in AI-driven healthcare and genomics initiatives are aimed at improving disease prediction, treatment, and patient care. **Israel:** Israel is making significant strides in AI and genomics, particularly in the field of cancer research. AI algorithms are being used to identify cancer biomarkers and develop innovative therapies. **Europe:** Various European countries, including Germany, France, and the Netherlands, are actively utilizing AI in genomics research. The European Union has launched projects like the European Genome-phenome Archive (EGA) to advance genomics through AI and data sharing.

India: India is increasingly adopting AI in genomics for applications such as disease risk assessment, pharmacogenomics, and drug discovery. The country's research institutions and biotech companies are contributing to global genomics efforts. **Australia:** Australia is investing in AI and genomics research to advance precision medicine and improve healthcare outcomes. Collaborations between academia and industry are driving innovations in the field. **South Korea:** South Korea is incorporating AI into genomics research to enhance cancer diagnosis and treatment.

The country is at the forefront of precision medicine initiatives involving AI-driven genomic analysis. These countries, along with many others, are actively benefiting from the synergy of AI and genomics, which has the potential to transform healthcare, accelerate scientific discoveries, and improve the understanding of genetic factors in health and disease on a global scale.

Future of AI in Genomic Biomarkers

Artificial intelligence (AI) is already having a major impact on genome biomarker research, and its importance is only going to grow in the future. AI can be used to analyze large and complex genomic datasets, identify patterns that would be difficult or impossible for humans to see, and make predictions about disease risk, prognosis, and response to treatment. It can be applied to discover new biomarkers. It can also be used to validate existing biomarkers and to determine their clinical utility more accurately. More precise AI-powered predictive models can be built to guide clinical decision-making in cancer and other diseases. AI is expected to play an even greater role in genome biomarker research to develop more comprehensive and accurate biomarkers. AI can be used to integrate data from multiple sources, such as genomics, proteomics, and metabolomics, to develop more comprehensive and accurate biomarkers. This could lead to the development of new biomarkers that can be used to diagnose diseases earlier and more accurately, to predict patient outcomes, and to guide personalized treatment decisions. AI can identify new drug targets by analyzing genomic data and identifying genes or other molecular targets that are involved in disease development and progression. This could lead to the development of new and more effective treatments for a wide range of diseases. It can make personalized medicine a reality by tailoring treatments to the individual patient's genetic makeup. This could lead to more effective and safer treatments for many different diseases. Overall, the future of AI in genome biomarker research is very bright. AI has the potential to revolutionize the way we diagnose, treat, and prevent diseases.

Challenges of AI in implementing genomic biomarkers

In the realm of AI methods, a significant challenge revolves around their interpretability. Unlike linear regression models, which offer a straightforward understanding of how covariates affect the dependent variable through linear relationships, data-driven machine learning models often appear as enigmatic "black boxes." Implementing genome biomarkers in the field of AI faces several

significant challenges. These challenges are crucial to understand and address to ensure the successful integration of AI into genomics research and personalized medicine. AI relies on large and high-quality datasets. Genomic data can be noisy, incomplete, and expensive to obtain. Ensuring the availability of diverse, well-annotated genomic data is essential for AI-driven biomarker discovery. AI models used for biomarker discovery often operate as "black boxes," making it challenging to understand the underlying biological mechanisms. Developing interpretable AI models is critical for gaining trust and insights from genomics data. Genomic datasets are typically high-dimensional, which can lead to overfitting when training AI models. Achieving robust generalization across diverse populations and datasets is a significant challenge. Genomic data is highly sensitive, and protecting individuals' privacy is paramount. AI solutions must address ethical concerns related to data sharing, consent, and the potential for unintended discrimination or misuse. Genomic analysis often requires massive computational resources, particularly for deep learning approaches. Ensuring access to powerful computing infrastructure can be a barrier for many research groups. Genetic variation among individuals and populations is substantial. AI models need to account for this heterogeneity to identify meaningful biomarkers that apply broadly. AI can identify associations in data, but it does not inherently understand the biology behind these associations.

Collaboration between AI experts and domain-specific biologists is crucial to translate AI findings into actionable insights. Biomarker candidates identified by AI must undergo rigorous validation to ensure their clinical relevance and reproducibility. This process is resource-intensive and time-consuming. Developing AI-driven biomarkers for clinical use requires navigating complex regulatory pathways. Ensuring compliance with regulatory standards and demonstrating safety and efficacy is a significant challenge. Integrating AI-driven biomarkers into clinical practice involves changes in healthcare workflows, physician training, and patient education. Overcoming these adoption barriers is vital for realizing the clinical impact of AI in genomics. Biased data can lead to biased AI models, potentially resulting in health disparities. Ensuring fairness in AI-driven biomarker discovery and addressing biases in data are essential considerations.

Genomic data often lacks longitudinal information, which is critical for understanding disease progression and treatment responses. AI methods need to accommodate dynamic data and account for temporal changes. Developing AI-driven biomarkers should consider cost-effectiveness, ensuring that the benefits outweigh the expenses associated with data acquisition, analysis, and

clinical implementation.

Addressing these challenges will require interdisciplinary collaboration, robust data governance, continuous validation, and ongoing research and development efforts to harness the full potential of AI in implementing genome biomarkers. The rapid proliferation of methodological variants and applications of AI methods in genomics leaves researchers grappling with a crucial question of how to select one algorithm over another or choose from related options. Addressing this conundrum is a burgeoning field known as automatic machine learning (AutoML). The objective of AutoML is to empower a computer algorithm to discern the most suitable algorithms and hyperparameters, thus eliminating the guesswork from the optimization process.

Conclusion

In conclusion, the future of AI in genomic biomarkers promises a paradigm shift in healthcare and biomedical research. AI-driven advancements hold the potential to revolutionize precision medicine, enabling early disease detection, tailored treatments, and more accurate risk assessments based on an individual's genetic profile. These developments are poised to enhance patient outcomes, accelerate drug discovery, and deepen our understanding of genetic contributions to health and disease. As AI continues to integrate with genomics, ethical considerations and regulatory frameworks will be paramount in ensuring responsible and equitable use of these transformative technologies.

COMPETING INTERESTS

The authors have no competing interests to declare.

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