Applications of AI in the Development of Personalized Medicine and Pharmacogenomics

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Abstract

As the applications of AI become more prominent in the healthcare industry, its relevance continues to grow in pharmacogenomics. Through AI methods like machine learning, scientists have been able to efficiently advance personalized drug development for patients with genetic variations and critical medical problems. This paper assesses how geneticists are using AI tools in pharmacogenomics to advance drug development in the healthcare industry. Pharmacogenomic testing has the potential to be a more efficient and cheaper alternative for traditional genetic testing; however, this testing is not currently used in primary health care due to a lack of knowledge on the functioning of pharmacogenomic tests. Additionally, many geneticists and the general public have shown skepticism surrounding the effects of AI and pharmacogenomic testing on other aspects of society – like environment and political concerns. This research introduces the basics of artificial intelligence and pharmacogenomics, later explaining how both of them are used together in the creation of medications and addressing public concerns surrounding the use of AI in pharmacogenomics. Despite these concerns, this research finds that AI is a helpful resource in genetic testing that will be of assistance to scientists and an ally in advancing drug development in healthcare.

Introduction

AI tools such as ChatGPT, MetaAI, and many others are increasingly popular around the world both for personal, business, and entertainment uses. AI is defined as the ability of machines to perform tasks that typically are done by human intelligence.[1] An uncommonly known evolving use of AI is its application in healthcare, specifically pharmacogenomics and drug development. Pharmacogenomics is a field of science that focuses on understanding whether the genetic differences between individuals can cause a difference in responses towards a certain drug or medication, while also being an approach for developing medications. Issues in the drug creation industry, like polypharmacy and time efficiency, are being improved through methods of pharmacogenomics. Polypharmacy is taking many medications simultaneously for a certain disease. This paper will analyze this evolution by questioning: How are geneticists using AI in pharmacogenomics to advance drug development in the healthcare industry and for the general public? What are the primary concerns of this novel technology? The biggest issue that has arisen with the advancement of pharmacogenomics and AI is how these two work hand in hand. Even with basic knowledge on the concept, many scientists and clinicians are not skilled in using the technology, especially pharmacogenomic tests. To combat this problem, this literature review discusses the methodology of pharmacogenomics and the different types of AI that assist the drug development process. The paper also addresses the negative aspects that are common in applications of AI, like environmental factors, political impacts, and race and gender divides.

With the evolving concept of artificial intelligence, the use of this technology in the healthcare industry was vastly researched. This was phase one: researching the different layers of biotechnology and the role of artificial intelligence in it. Pharmacogenomics was found to be a prevalent topic. Pharmacogenomics is a genetic analysis technique for drug development that uses artificial intelligence to create personalized medication for patients based on their genetic variation. As my research continued, there was a limited amount of peer reviewed data about pharmacogenomics and AI found.

Section 1: How is AI used in Pharmacogenomics

Pharmacogenomics is a field of science that focuses on understanding whether the genetic differences between individuals cause a difference in responses towards a certain drug or medication, while also being a guideline for developing medications.[2] The goal of scientists using pharmacogenomics in the healthcare industry is to eventually create the system of personalized drugs.The idea of personalized medicine stems from the concept of medicine affecting people differently, depending on their age, gender, drug reactions, and heritable traitsThe basic process of pharmacogenomics starts with sample collection of patients' data, including their genetic data, familial history, and medical history. From there, analysis and genotyping of data occurs, which is used to connect the genetic components to human diseases. The analysis of genetic data has been linked with AI, where machine learning scans medical records to identify the genetic variations in an individual. This speeds up the process of the implementation of pharmacogenomics.[3] All together, pharmacogenomics is used by scientists with the aim of making the process of drug development more accurate and efficient through the help of AI.

In the healthcare industry, AI has the potential to help scientists diagnose disease, helping healthcare workers in patient care analyze large amounts of data. Specifically, within the applications of AI, machine learning is one of the most prevalent in executing tasks related to pharmacogenomics. Machine learning is the technique of fitting models into data and training models with data so that algorithms can learn and execute tasks without instructions.[4] For large amounts of genetic data and clinical data, machine learning is able to analyze the data and find patterns within the information that a human wasn't able to point out.[5] Within pharmacogenomics, the process of machine learning is able to predict what treatments are likely to be the most beneficial for a patient based on their genetic information and specific attributes.[6]

There are two types of machine learning that have applications within the medical industry: supervised and unsupervised learning. Supervised learning is when algorithms are trained on labeled datasets to form predictions, like an automated detection of a genetic variation within a DNA strand. On the other hand, unsupervised machine learning is when algorithms make predictions with undetermined labels, like clustering patients' health records based on a certain similarity. In pharmacogenomics,supervised learning is more beneficial due to the accuracy of machine learning analyzing large amounts of genetic data.[7] In the last decade, machine learning has allowed AI systems to predict outcomes and plan medical data.[8]

Microarrays are laboratory tools part of artificial intelligence that are used in pharmacogenomics. Microarrays are a general laboratory approach in pharmacogenomics that run a genetic analysis of an organism for diseases that have been standardized. These microarrays are able to detect multiple pathogens, or organisms that cause disease, in the DNA simultaneously. Some aspects that microarrays detect to determine the diseases are DNA content, host gene products, and more, which allows for this technology to discriminate between various models of a specific disease.[9]

In the medical industry, a huge issue is the cost, ineffectiveness, and creation of drugs for many diseases. Before a drug is introduced to the market, scientists have to work in laboratories for clinical testing where the drugs are examined to see whether they match the regulatory standards of medicine.a majority of drugs fail, which results in a loss of money, time, and effort. Along with this, most diseases that drugs are created on, for example cancer and Alzheimer's disease, include polygenic traits, meaning that genetics play a role in the disease.a bioanalytical platform can provide clinical support to increase medication management in the primary healthcare industry.[10] To support the use of pharmacogenomics within the primary healthcare industry, scientists discuss the issue of polypharmacy, or the use of multiple drugs by a patient, to describe how many patients go through unwanted reactions to drugs and hepatic cytochrome enzyme-mediated drug-drug interactions. Polypharmacy also causes an increase in costs within healthcare for patients. To prevent these side effects, t pharmacogenomics can inform scientists about human genetic variations and how they correlate to drug usage and diseases. AI comes into play during the data analysis phase when geneticists run AI to make it analyze the symptoms and genetics of humans within medical records. The authors conclude that their vision for pharmacogenomics is one that is currently impractical due to it not being accessible to primary health care, but if it is proven that AI and pharmacogenomics are cost-effective and result in better health outcomes, it can be beneficial for the future of drug creation.

An example of the application of AI in pharmacogenomics can be seen at Texas A&M Family Medicine Program, where scientists are studying chronic disease. Firstly, genomicists begin by studying medical records which contain basic medical and family information, which is the foundation of actual pharmacogenomic testing. From there, more information about the patient is gathered by clinicians through physical exams and blood tests which get processed by AI tools in a bioanalytical laboratory. Then, if there are any symptoms seen in the information that are suggestive of chronic disease, alternative medication is recommended by the physician to the patient. This step is where AI comes into play; Pharcognemomics uses artificial intelligence to analyze the symptoms reported in the medical reports that might be worrisome. These symptoms include side effects of medications or the development of a past or present diagnosis. This analysis then plays a crucial role in helping geneticists develop a model for the predictions of how a disease will impact a patient, which is an aid for clinicians. The process of

AI and pharmacogenomics takes less time to complete tasks that would usually take long periods by a human, while simultaneously helping many patients provide accurate analysis and results.[11]

A review of novel AI techniques and tools in the field of pharmacogenomics shows AI to be most useful in the processing of genetic data and analysis of medical data. With the immense amounts of data that are presented to scientists and geneticists for personalized drug development, methods of machine learning make the process of data analysis efficient by using algorithms compared to traditional linear regression methods. Along with time efficiency, AI tools such as machine learning also provide accurate results, which supports the credibility of these resources. AI tools are being used to assist scientists by detecting genetic variations in large amounts of genetic data, which in turn is assisting the endeavor of drug development.

Section 2: Impact of AI and Pharmacogenomics

Compared to standard testing for patients, pharmacogenomics tests (PGx) have proven to be more cost effective by scientists. Most pharmacogenomic tests were cost-effective with the decrease in PGx prices. 77% of the articles that were studied in the analysis were considered as cost-effective, 22 out of 23 clopidogrel tests being determined as cost-effective. A factor of the cost impact is based on whether the tests are single-gene or multigene tests. The costs were compared between a multigene antiplatelet therapy test and a single gene test for pain treatment, with the conclusion that the multigene test had a more favorable incremental cost-effectiveness ratio.[12]

However, with the cost-effectiveness of PGx testing comes the concern of racial and gender inequality in the process of testing. In order to analyze the racial bias in pharmacogenomic testing, scientist Manuel Corpus and peers (2024) looked into the biogeographical data of individual patients in PharmGKB, a resource in embedding pharmacogenomics data. The researchers found around 509 biogeographical groups from the data, most people being considered as heterogeneous. Heterogeneity is someone having a combination of multiple ancestry groups. These groups were disregarded from the study due to the difficulty in categorizing the ancestry groups, which was a potential issue in selection bias against race. The underrepresented racial groups were determined to be Africans, Latinos, and indigenous Americans, while the prominent racial group found was European. There has also been a lack of data for women For example, in the testing of certain diseases such as chronic kidney disease, out of the tested individuals, only 44% were women, even when they were more likely to get the disease. This type of bias is also seen in the study of effects of drugs on individuals.[13]

With the rise of AI in pharmacogenomics, scientists are concerned about AI in the healthcare industry, including pharmacogenomics. Large businesses control the production of technology that is used everyday, like Google, Apple, and Microsoft. In pharmacogenomics, Thermo Fisher Scientifics uses AI in genetic testing.[14] According to researchers Luitse and Denkena, large language models (LLMs) are data processing models that are able to understand and generate human text based on large data sets.[15] They raise the concern that the trend of LLMs in natural learning processes causes a risk of monopolization and increasing dependence on high-scale AI companies. AI machines, like Transformer architecture – a neural network architecture that transforms an input sequence to an output sequence – requires the usage of parameters, which are variables optimized to minimize error to display that the more the parameters the more effectively data is handled. A table shows this trend in relation to the different versions of Transformer technology, while also showing that these models are controlled by highly industrialized companies who have the resources to continue the AI technology, like Google, OpenAI, and more.[16] AI requires large amounts of energy to execute tasks, and this is only possible when companies who have access to an abundance of resources control the sector of AI.

With the use of energy and power to perform tasks by AI, there is concern about AI's environmental impact and consumption of natural resources. One main resource that is being used in aspects of AI and data creation is water. Water is used in two ways: it is used for cooling data centers and the production of microchips for AI. During the training processes of many AI models, hundred thousands of liters of water are required for an AI model that is equivalent to the human brain processing, a study estimating around 126,000 liters.[17] As AI models become more used and advanced, the consumption of water by this technology will also increase, having a greater environmental impact.

Limitations

Pharmacogenomics is a field of biotechnology that is advancing everyday and is comparatively new to the realm of artificial intelligence. This created an issue in the research process because many answers to specific questions were not yet reflected in the published record, like finding current statistics on the utilization of pharmacogenomic tests. Along with this, the complexity of pharmacogenomics caused there to be an influx of different terms being thrown around that had minimal information about them, causing understanding of the topic to get distracted by other information. The topic of pharmacogenomics has many gaps due to the newness of the concepts, also making it a divisive field in which there were constant reminders to focus on the main topics of pharmacogenomics. This research paper drew on the recent works of Silva (2021), Morris (2022) and their examination on the role of AI in advancing the field of pharmacogenomics. As this paper was being finalized, Taherdoost and Ghofrani (2024) published their paper titled, "AI's role in revolutionizing personalized medicine by reshaping pharmacogenomics and drug therapy." While relevant to this research, there wasn't enough time to consult the article, though the findings of this paper resonate with the arguments of this paper, and are largely in consensus.

Conclusion

In the last few years, Artificial intelligence is commonly used in the world around us through common resources such as ChatGPT, and is seen through social media and applications in schools. A new application of AI is seen in a sector of healthcare: Pharmacogenomics, a field of science that determines how the genetic differences between individuals impact the way in which a certain drug or medication affects the health of a patient. Some common issues that pharmacogenomics is being used to solve are polypharmacy and the efficiency of drug creation, in harmony with AI methods to analyze genetic data. The combination of AI and pharmacogenomics brought up speculation how geneticists were using AI applications in personalized medicine and pharmacogenomics, along with which technology was being used. Through different AI tools, such as machine learning, pharmacogenomic tests are able to identify genetic differences in an individual to create personalized drugs. However, the application of AI in pharmacogenomics also has its drawbacks, consisting of environmental issues, political concerns, and gender and racial inequality caused by the systems of pharmacogenomic tests. Through this all, the applications of AI prove to be beneficial to the process of pharmacogenomics, by making it an efficient and manageable resource for geneticists to use in the operation of drug development.

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