Barry P Chaiken, MD June 8, 2023

Unleashing Precision Medicine to Deliver Personalized Care

Precision medicine, also known as personalized medicine, is a modern approach to healthcare that considers individual variability in genes, environment, and lifestyle for each person.

This approach allows doctors and researchers to predict more accurately which treatment and prevention strategies for a particular disease will work in which groups of people. It contrasts with a one-size-fits-all approach, where disease treatment and prevention strategies address the average patient with less consideration for individual differences.

At the core of precision medicine is tailoring treatment to each patient's characteristics. This involves understanding how a person's genetic makeup, environment, and lifestyle can impact their health and risk of disease. Understanding these factors allows us to design more effective treatments tailored to patients' needs.

For example, in oncology, precision medicine might involve targeted therapies designed to treat specific types of cancer cells based on their genetic characteristics. This approach can lead to more effective treatments with fewer side effects than traditional chemotherapy.

Precision medicine represents a significant shift in our approach to health care, from a reactive system that responds to disease to a proactive system that aims to prevent illness and maintain health. It is a promising field that has the potential to revolutionize the way we understand and treat disease.

Practice Guidelines vs. Precision Medicine

Practice guidelines used in medicine today are typically based on evidence from large-scale clinical trials. These guidelines provide a general framework for treating patients with a specific condition and are designed to apply to the "average" patient. They are extremely valuable in standardizing care and ensuring that patients receive treatments that have been proven effective

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in large groups of people. However, these guidelines often do not consider individual variability among patients.

Precision medicine, on the other hand, aims to tailor medical treatment to the individual characteristics of each patient. It acknowledges that disease is often a complex interplay of genetic, environmental, and lifestyle factors and that each patient's combination of these factors is unique. Therefore, there may be more effective and efficient ways to treat every patient than traditional practice guidelines "one-size-fits-all" approach.

For example, in the context of cancer treatment, practice guidelines might recommend a specific chemotherapy regimen for all patients with a particular type of cancer. However, precision medicine might reveal that a subset of these patients has a specific genetic mutation that makes their cancer cells resistant to this chemotherapy but susceptible to a different drug. In this case, there are better courses than following the practice guidelines for these patients.

In essence, while practice guidelines aim to standardize care and reduce variability in treatment, precision medicine seeks to embrace this variability and use it to tailor therapy to individual patients. Both approaches have their place in modern medicine, and the best care often involves a combination of the two.

Precision Asthma Treatment

Asthma is a chronic disease characterized by inflammation and airway narrowing, leading to wheezing, coughing, and shortness of breath. However, not all asthma is the same. Various factors, including allergens, exercise, infections, and even stress, can trigger asthma, and its severity and response to treatment can vary widely between individuals.

Traditionally, clinicians manage asthma with a combination of quick-relief medications for acute symptoms and long-term control medicines to reduce inflammation and prevent asthma attacks. However, these treatments are not always effective for all patients, and some patients may experience significant side effects.

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Precision medicine is helping to improve asthma treatment by identifying different asthma subtypes, or "endotypes," based on their underlying biological mechanisms. For example, some people have "allergic asthma," which is triggered by allergens and associated with high levels of a type of white blood cell called eosinophils. Other people have "non-allergic asthma," which is not associated with allergens or eosinophils and may be triggered by factors like exercise or stress.

These different endotypes can respond differently to different treatments. For example, a group of drugs called biologics, which target specific immune pathways, have been developed to treat severe asthma. Some of these drugs are specifically designed to reduce eosinophil levels and are particularly effective for treating allergic asthma. However, they may not be effective for treating non-allergic asthma.

Doctors can better tailor their treatment by identifying a patient's asthma endotype through blood or sputum tests. For example, a patient with allergic asthma might be prescribed a biologic targeting eosinophils, while a non-allergic one might be prescribed a different treatment.

In this way, precision medicine is helping to move asthma treatment away from a one-size-fits-all approach and towards a more personalized approach that can improve outcomes and reduce side effects for patients.

Precision Medicine Challenges

While precision medicine holds great promise, there are several obstacles to its more comprehensive implementation:

1. Data Collection and Analysis: Precision medicine relies on the collection and analysis of large amounts of data, including genetic data, environmental data, lifestyle data, and more. Collecting this data can be time-consuming and expensive, and analyzing it requires sophisticated computational tools and expertise.

2. Interpretation of Genetic Information: While we have made great strides in our ability to sequence genomes, our understanding of what genetic variations mean still needs improvement. Many genetic variations have unknown significance, and it can be challenging to determine whether

What is Hallucination in AI?

Hallucination in AI refers to the generation of outputs that may sound plausible but are either factually incorrect or unrelated to the given context. These outputs often emerge from the AI model's inherent biases, lack of real-world understanding, or training data limitations. In other words, the AI system "hallucinates" information that it has not been explicitly trained on, leading to unreliable or misleading responses.

Source: https://bernardmarr.com/chatgpt-what-are-hallucinations-and-why-are-they-a-problem-for-ai-systems/

a particular variation is causing a disease or is just a harmless difference.

3. Privacy and Ethical Concerns: Collecting and using personal health data, including genetic data, raises important Here are some references that you may find helpful in understanding these models.

4. Healthcare Infrastructure: Many healthcare systems require increased capabilities to incorporate precision medicine approaches. This includes practical issues like the need for electronic health records (EHRs) to ingest genetic and other data and more fundamental matters like training healthcare providers in genomics and precision medicine.

5. Cost and Accessibility: Many precision medicine approaches, such as genetic testing and targeted therapies, are expensive. There are concerns that the benefits of precision medicine may not be accessible to all patients, leading to disparities in care.

6. Regulatory Challenges: Regulatory frameworks for approving and using precision medicine treatments, particularly those based on genetic information, are still being developed and can be complex.

Despite these challenges, progress is being made in these areas, and the potential benefits of precision medicine make it a worthwhile pursuit. It is an exciting field that will likely be increasingly important in healthcare in the coming years.

Precision Medicine Challenges

EHRs are crucial in delivering precision medicine, acting as a double-edged sword with benefits and challenges. On the one hand, EHRs serve as a powerful tool for data aggregation, providing a comprehensive view of a patient's medical history, including diagnoses, treatments, outcomes, demographic information, lifestyle factors, and potentially genetic data. This wealth of information can significantly aid physicians in making more personalized treatment decisions.

Moreover, EHRs facilitate the sharing of data between different healthcare providers, specialists, and institutions, which is particularly important for precision medicine that often involves multidisciplinary care teams.

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Additionally, EHRs can incorporate clinical decision support tools that provide physicians with evidence-based treatment recommendations tailored to individual patients, integrating genetic and other data to offer precision medicine recommendations. From a research perspective, the aggregated EHR data can be a treasure trove for discovering new disease markers, understanding treatment responses, and gaining other insights that can drive precision medicine forward.

On the flip side, EHRs present several challenges. The quality and standardization of EHR data can be inconsistent, with different healthcare providers using different terms and codes for the same conditions or treatments, making it challenging to accurately aggregate and analyze EHR data for precision medicine. Interoperability between different EHR systems is often limited, hindering data sharing between healthcare providers and institutions.

Another significant challenge is the integration of genomic data. EHRs were not originally designed to handle the large amounts of complex genomic data often involved in precision medicine, and integrating and interpreting this data in a clinically useful way can be challenging. Lastly, using EHR data for precision medicine raises essential privacy and security concerns. Patients must be assured that their data will be kept secure and used responsibly.

Role of AI

Artificial Intelligence (AI) holds significant potential in advancing the field of precision medicine. It can help in various ways, from analyzing large datasets to predicting patient outcomes and personalizing treatment plans.

One of the critical applications of AI in precision medicine is the analysis of large and complex datasets. Precision medicine often uses genomics, proteomics, and other "omics" technologies that generate vast amounts of data. AI, particularly machine learning algorithms, can help analyze these data to identify patterns and correlations that traditional statistical methods might miss. For instance, AI can help identify genetic variants associated with disease risk or treatment response, which can guide the development of personalized treatment plans.

AI can also play a significant role in imaging analysis. In radiology, pathology, and other imaging-based disciplines, AI can help analyze images to identify disease markers that might be too subtle for the human eye to detect. This can aid in early

Navigating the Code

The healthcare industry, unlike many others, runs on time-tested ways to practice excellence in medicine. But does that mean adherence to practices and processes that are fifty, seventy, even a hundred years old?

Dr. Barry P. Chaiken thinks not. His 25+ years of experience as a physician and an informaticist, he believes information technology is healthcare's greatest problem-solving tool for resolving the greatest medical and business problems of the 21st century.

disease detection, prognosis, and monitoring of treatment response.

Furthermore, AI-powered predictive modeling can help forecast patient outcomes based on their unique characteristics. For example, AI algorithms can predict which patients are at high risk of disease complications or recurrence, allowing for early intervention and personalized care management.

AI can also support clinical decision-making. AI-powered clinical decision support systems can provide physicians personalized treatment recommendations based on a patient's unique genetic, clinical, and lifestyle data. These systems can also incorporate the latest research findings, helping to bridge the gap between research and clinical practice.

Lastly, AI can help in drug discovery and development, a critical aspect of precision medicine. AI can expedite the process of identifying potential therapeutic targets and predicting drug efficacy and safety, thereby aiding in the development of personalized therapies.

However, it's important to note that while AI holds great promise, it also presents challenges, including data privacy and security, algorithmic bias, and the need for transparency and interpretability in AI decision-making. Addressing these challenges is crucial to ensure AI's responsible and effective use in precision medicine.

In conclusion, precision medicine holds great promise for the future of healthcare, offering a shift away from the application of population-based treatment guidelines to a more personalized one. It is a medical model that proposes the customization of healthcare, with decisions and treatments tailored to individual patients.

Precision medicine has the potential to revolutionize healthcare by providing more personalized and effective treatments. It is a promising field that is likely to grow and evolve as our understanding of diseases and their underlying mechanisms continues to improve.

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Author Note: I wrote this article using ChatGPT (4.0). By requesting several "regenerations" of the responses, I constructed a more informative article from pieces of each version. This is the finished document.